BioLaw Journal
Rivista di BioDiritto

1 21

Special Issue

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Special Issue || Law, Genetics and Genomics: An Unfolding Relationship

The online Journal about law and life sciences

BioLaw Journal – Rivista di BioDiritto

Email: biodiritto@gmail.org
Website: rivista.biodiritto.org/ojs

Peer review system: All academic articles that are submitted to *BioLaw Journal – Rivista di BioDiritto* are subject to a double blind peer review. Referees remain anonymous for the author during the review procedure and the author's name is removed from the manuscript under review.

May 2021
ISSN 2284-4503

UNIVERSITY OF TRENTO - Italy

Università degli Studi di Trento
Via Calepina, 14 – 38122 Trento
Registrazione presso il Tribunale di Trento n. 6 dell'11/04/2014

In collaborazione con

BioDiritto
UNIVERSITÀ DI PARMA
DIPARTIMENTO DI GIURISPRUDENZA, STUDI POLITICI E INTERNAZIONALI
UCB University Center for Bioethics
FONDAZIONE LEONARDO CIVILTÀ delle MACCHINE

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Cover design: Marta Tomasi

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BioLaw Journal – Rivista di BioDiritto
Special issue n. 1/2021

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Law, Genetics and Genomics: An Unfolding Relationship

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The relationship between genetics/genomics and the legal and ethical landscape has been long and varied, and it is continually evolving. Depending on the moment in history and the specific area involved, this relationship has proven tragic (consider eugenics and the laws it inspired), beneficial (consider the many criminal cases solved through forensic DNA) and promising (consider attempts to regulate emerging gene editing techniques) – but it has never been easy.

The idea to dedicate a Special issue of *BioLaw Journal* to the origins, state of the art and future of this interrelation emerged during an international symposium organised by the Italian Chapter of the International Society of Public Law (ICON-S) in Florence in 2019, addressing the topic of *New Technologies and the Future of Public Law*. A stimulating discussion on the new challenges of genome editing convinced us that trying to bridge between past and present was important in order to better imagine the future.

Aware of this issue's challenging and multifaceted nature, we involved outstanding experts who had devoted years of their research activities to these topics, together with passionate young scholars whose imaginations had more recently been captured by these complexities. Relying on a cross-sectoral approach that embraced – beyond law – medicine, the history of medicine, philosophy and bioethics, we cooperated with interested Authors from different parts of the world (beyond Italy, including Australia, Belgium, Canada, China, France, Hungary, the United Kingdom, Portugal, Qatar, South Africa, Spain, Sweden and the United States), showcasing the global dimension of genetics challenges.

Far from encapsulating the essential features of human nature, genetics is undoubtedly tackling the origins and future development of the very idea of humanity, rooted in history and prospects for evolution. From this consideration, we decided to organise this Special issue on the basis of a timeline moving from traditional concerns towards the most innovative and unexplored issues.

To prepare the field for this discussion, the first part of this Special issue comprises a Forum in which experts share some visions of the most sensitive matters. The points touched upon in this first part of the issue mirror the broader reflections presented thereafter, offering a precious insight into the remainder of the Issue.

The debate is opened by three scientists (Simonato, Verlengia and Barbujani) who offer critical understandings of the potential and risks of genetic knowledge as a complex subject – and of the indispensable need for interdisciplinary debate on its applications.

The following contributions discuss the specificities of genetic information and the dynamics it triggers in different fields. Genetic data's ultra-individual nature, for instance, requires an in-depth consideration of the importance of sharing within the so called "biological group" (Rial-

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Sebbag). Beyond the clinical setting, sharing also occurs at a different level in the scientific research field, revealing tensions between the "open data agenda" and the protection of traditional privacy standards and confidentiality, along with an ideal of self-determination. These tensions are particularly evident in specific areas of research, such as research on rare diseases (Rothstein), in which a lack of affected individuals to study may compromise research strategies, and shared research protocols are needed beyond different national systems. More generally, the frictions produced by the global dimension of research require the law to rethink itself – sometimes in provocative terms – in order to reconsider its role and assess the importance of spaces dedicated to different instruments, such as sources of soft law or mechanisms of trust and responsibility based, for example, on combined professional discretion and accountability requirements, along with the supervisory role played by institutions such as Institutional Review Boards or ethic committees (Knoppers and Beauvais, Rothstein). An area where these needs are particularly evident is in the application of new gene editing technologies, where procedural questions about how to deliberate and make regulatory decisions play a crucial role (Casabona).

The section closes with a forward-looking philosophical reflection on the concept of *embodiment* between the human and the artificial, with which moving from a biological inheritance to a machine-based future is not a neutral process but, rather, a cultural inheritance of modernity (Calo).

The same timeline that characterises the introductory Forum structures the following part, made of two Sections of essays: "Troubling historical roots and contemporary challenges" and "Views into the future".

The first Section progresses from the history of genetics (Ampollini) and the dramatic consequences of its misinterpretations of contents by public authorities (Tomasi) to the current challenges stemming from the field's actual characteristics. In this section, notions of *autonomy* and *privacy*, already presented in the introductory section, re-emerge in specific declinations. First, some reflections are devoted to the clinical field, within which a constitutionally oriented vision must be applied. Accordingly, the concept of *self-determination* and the related instrument of *consent* must confront the fact that a person cannot be understood as an atomistic entity, disconnected from the relationships in which he/she lives. For instance, the configuration of a right not to know "incidental findings", is articulated not only in terms of *self-determination* but also *solidarity* and *responsibility* (Cozzi). Similarly, the field of newborn screenings' regulation presents an urgent need for balance between protecting children's best interests, exercising parental responsibility and the collective interest in preventing serious diseases whose management strongly influences – from a socio-economic perspective – the whole community (Di Costanzo). Moreover, these complex balances and their resulting conflicts find different points of synthesis, depending on their surrounding conditions, which can determine – as in the case of the current Coronavirus disease pandemic – exceptional states that subvert traditional logics (Brownsword and Wale).

The context in which these issues primarily emerge is family relationships, where genetic bonds produce a strong impact. Two papers in this Special issue are devoted to a search for origins. The first of these papers (Agosta) compares the case laws of the European Court of Human Rights and Italian Constitutional Court with regard to cases concerning the search for a

mother's identity. The second of these papers (Busatta and Penasa) is dedicated to the right to know one's genetic origin, its profoundly relational nature and the need to balance it with other competing individual rights, which – according to the concrete context at stake (anonymous birth, gamete donation and surrogacy) – can be traced back to donors, to gestational mothers and even to children themselves (i.e. the right to a safe birth, as in the case of anonymous birth). Other familial issues arising from genetic data shared nature concern the need to communicate potentially relevant information to individuals with a qualified interest. In this context, any processing of genetic data (also under the European General Data Protection Regulation) should be regarded as an exercise of balancing interests which cannot be limited to overprotecting the right to self-determination over data (De Miguel Beriain and Jove).

Very similar approaches, which require overcoming a purely individualistic logic of "closedness", also emerge in different fields, such as scientific research. This development shows that genetic testing is a "complex and integrated enterprise" which management requires to reinterpret and sometimes transcend existing regulatory instruments (Slokenberga). Samples and data are the most valuable resources to the aims of genetic research, and they are more and more often collected from outside traditional research settings (Shabani), thanks to various online tools and platforms (e.g. electronic health records, mobile health apps, disease registries and patient-generated databases). These instruments, together with the important wide sharing of research materials beyond national borders and at different times, require an in-depth discussion of the challenges associated with data processing's governance and traditional instruments of self-determination, such as informed consent. As the two examples presented in this part of the Special issue show – one related to a specific national context (Mahomed and Staunton) and the other related to a concrete research project in South Tyrol (Biasiotto, Pramstaller and Mascalzoni) – privacy itself cannot protect participants or properly balance all relevant interests. Especially when the sharing and reuse of samples and data are involved, privacy must be combined with external governance mechanisms to sustain and promote trust and ensure transparency. Not only are privacy and consent under pressure, but so are intellectual property rights; for this reason, the Special issue is enriched by a reflection that assesses possible scenarios for biotech-related patent applications' future, starting with a case-law analysis (Lucchi).

The contents of this Special issue also focus on the use of genetic analysis in the field of criminal investigations. Beyond the problems raised by criminal lawyers, the contributions collected here reflect on the elements emphasised in clinical and scientific research from a different perspective: the links within the family group, which allow the development of sophisticated investigative techniques, from "familial searching" to "forensic genetic genealogy" (Formici), and the need to exchange samples and data at the international level – in this case, for justice purposes (Scaffardi). The challenge is to strike a proper balance between the public interest in security and a rapid, efficient identification of unknown offenders, as well as the dangerous shift towards "genetic surveillance" which jeopardises individual rights.

The issue's second Section, dedicated to the "Views into the future", considers genetics' ability to shape ourselves and the world around us. The first contribution considers the difficulties of tracing a sharp line between what is "natural"

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and what is "synthetic" and the importance of general principles, such as dignity and justice, as well as specific instruments, such as informed consent, which cannot be fully renounced ahead of technological advancements of bioengineering and their circulation in a free market (Đuković). Among the various technologies that can now affect the concept of "naturalness", a wide-ranging ethical and legal debate surrounds gene editing techniques. In particular, genetic engineering is on the cusp of a transformational change, thanks to CRISPR-Cas9, a tool that allows scientists to alter different organisms' DNA – including humans – with high speed and precision and at relatively low costs. Two papers in this Issue are specifically dedicated to these techniques and particularly address the importance of a global approach to this topic, which can rely on proper regulation levels, based on human rights law and international law principles (Poli), and on the recourse to innovative instruments, such as deliberative democracy (Ragone). The rising chance of modifying genetic features or selecting individuals on the basis of their genetic makeup – resulting from advances in genetic knowledge – affect parent-child relationships, sparking arguments about intergenerational justice. Regarding these sensitive issues, diverse attitudes and opinions can be shown. The views presented in two of the papers collected in this Special issue seem to occupy the two extremes of this spectrum, but they are actually inextricably linked. On the one hand, one contribution addresses parents' civil liability *vis-à-vis* claims presented by their children, not only for their actions but also for their omissions (Payán Ellacuria). On the other hand, a strong position favouring gamete selection in order to allow parents to choose non-health-related features is presented and discussed (Raposo).

We decided to place two very different papers sharing a common "take-home" message at the end of this Special issue. This message can be summarised as the importance of a systemic vision in addressing the regulations of genetic/genomic applications. Legal and ethical reflections must understand that no intervention or activity in the realm of genetics can be considered neutral – that interrelations are a fundamental feature of our world. Even choices concerning the smallest insect inhabiting our planet must be carefully weighed because they entail broader considerations that affect all living beings and our entire environment (Annoni and Pievani). Finally, human beings today cannot be considered solitary agents: our biological niche, built on a complex network of relations, requires a consideration of not only other (past, present and future) human beings and animals but also machines (Santosuosso), always expanding our responsibilities.

The picture is inevitably incomplete, but the papers collected in this Special issue offer vivid insights into continually important issues surrounding genetic knowledge – both within and beyond ethico-legal debates.

We thank each contributing Author for their commitment, essential contribution, patience and enthusiasm for this discussion, which aimed to facilitate intergenerational and interdisciplinary encounters. Special thanks are also due to Elena Scalcon and Federica Fortunato, whose assistance and expertise were essential for this Issue's accurate editing.

BioLaw Journal is very attached to the vision of (bio)law that emerges from this collective effort. This law pays attention to the concrete reality which it aims to regulate, and it is up-to-date. It is proportionate, non-invasive, plural and non-value oriented. Finally, it recognises the past

which it encapsulates, and it can adapt to the challenges ahead.

ISSN 2284-4503

Law, genes and bioethics: A biomedical perspective

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1. Introduction: what is a complex trait?

"We wish to suggest a structure for the salt of deoxyribose nucleic acid (D.N.A.). This structure has novel features which are of considerable biological interest"[1]. This statement by Nobel Prize winners James Watson and Francis Crick sparked the first genetics revolution in 1953. Fifty years later, with the completion of the human genome project, a second genetic revolution took place with the publication of the first draft mapping the complete human genome sequence[2]. With time, errors in the first draft were corrected and a swift flourishing of powerful technologies are nowadays enabling the sequencing of many thousands of human genomes per year, a number that is constantly growing. "We used to think that our fate was in our stars, but now we know that, in large measure, our fate is in our genes" stated James Watson at the start of the Human Genome project. Now, twenty years later, he would probably admit that the project fell short in keeping these promises, for two reasons. First, our fate is much too complicated to be all written in our DNA; second, whatever might be written in the DNA, reading it is just the first step of a much more complex task we have not yet accomplished, that is, to understand it.

Our genome is often metaphorically described as a text. The genome has an alphabet, the four bases A, C, G and T forming the 46 long "books" (our chromosomes) where the text is contained. And it has a lexicon, 21.000 or so words or genes[3], i.e. the regions of the genome that are transcribed into RNA and then translated to create proteins. Knowledge of these genes already allows us to diagnose many genetic diseases, including muscular dystrophies, hemophilia and cystic fibrosis, all disorders that are caused by the malfunctioning of a single gene.

However, we do not yet understand the genome syntax. The diseases causing the heaviest health burden (like cancers, cardiovascular and neurodegenerative disorders) depend on multiple interactions among many genetic and environmental factors, i.e. are complex traits that do not behave according to simple Mendelian inheritance laws. Because the genes involved in these diseases are many, each playing a limited role, their identification and use to estimate disease risk has proved difficult. While we are far from achieving a general, clear understanding of the genetic bases of complex diseases, even more

[1] J.D. WATSON, F.H.C. CRICK, *Molecular structure of nucleic acids: A structure for deoxyribose nucleic acid*, in *Nature*, 171, 4356, 1953, 737-738.

[2] International Human Genome Sequencing Consortium, *Initial sequencing and analysis of the human genome*, in *Nature*, 409, 2001, 860-921; J.C. VENTER et al., *The sequence of the human genome,* in *Science*, 291, 2001, 1304–1351.

[3] D.R. ZERBINO, A. FRANKISH, P. FLICEK, *Progress, challenges, and surprises in annotating the human genome,* in *Annual Review of Genomics and Human Genetics*, 21, 2020, 55-79.

incomplete is our understanding of the genetic bases of non-pathological complex traits, such as those related to personality, cognitive abilities or emotions. These traits do recur in families, and hence there is reason to suspect that genes play a role. However, it is notoriously hard to dissect the effects of shared genes from those of shared environments. One example of a particularly unsuccessful attempt to identify the genes governing a complex trait has been the search for IQ (Intelligence Quotient) determining-genes. A recent, colossal study of nearly 80,000 subjects led to the identification of 22 candidate genes, which, however, could globally account for just 1,5% of the differences between very high and very low IQ values[4].

While science struggles to address these issues, the advancement in knowledge already poses many outstanding ethical and legal problems. Below, we discuss some of these issues.

2. Criminality genes

Despite all, deterministic views have all but disappeared in biology, and are well documented by the century-long search for genes determining criminal attitudes. Much like intelligence, criminal behavior is another ill-defined topic, and so it comes as no surprise that the investigation of criminality genes has been as frustrating and vain as the search of intelligence genes. Around 1960, the idea that an extra Y-chromosome in males could lead to criminal behavior achieved popularity, largely through the media. The first case of a man carrying an XYY chromosome complement was described in 1961, and four years later Patricia Jacobs published a survey of 315 men at a hospital for developmentally disabled in Scotland, 9 of whom (all characterized as violent criminals) apparently carrying an extra Y chromosome[5]. In 1968, a US serial killer, Richard Speck, was described as XYY in three articles on the New York Times. In fact, Speck had a normal chromosome set, but this fake news contributed to create the myth of the XYY man as a congenital criminal, which persisted even when M. Court Brown failed to confirm it in a large study (more than 5000 subjects) in Scottish prisons[6]. The Y chromosome is a small chromosome; the simplest explanation for Jacobs' findings was that she mistook for Y chromosomes some dark spots on the printed photographs. Yet, as late as 1974, 13 men and boys with XYY chromosome complement were sentenced to chemical castration in Maryland[7]. The scientifically unsupported stereotype of XYY men as violent criminals lasted for decades after its scientific dismissal; it was used as a plot device in horror films such as Dario Argento's *The Cat o' Nine Tails* (1971) and David Fincher's *Alien 3* (1992).

In later times, another gene, *MAOA*, mapping on the X-chromosome, enjoyed a transient popularity as a criminality gene. *MAOA* codes for a protein, monoamine oxidase A, involved in the metabolism of several neurotransmitters, such as dopamine and serotonin, and hence is a key regulator of many functions of the brain. A low-activity variant, *MAOA-L*, was identified in a Dutch family in which several males had shown borderline mental retardation and abnormal behavior

[4] S. Sniekers et al., *Genome-wide association meta-analysis of 78,308 individuals identifies new loci and genes influencing human intelligence,* in *Nature Genetics,* 49, 2017, 1107-1112.

[5] P. Jacobs et al., *Aggressive behavior, mental sub-normality and the XYY male,* in *Nature*, 208, 1965, 1351-1352.

[6] M. Court Brown, *Males with an XYY sex chromosome complement,* in *Journal of Medical Genetics*, 5, 1968, 341-359.

[7] R. Pyeritz et al., *The XYY male: The making of a myth,* in Ann Arbor Science for the People Collective (eds.) *Biology as a social weapon,* 1977, 86-100.

including impulsive aggression, arson, attempted rape, and exhibitionism[8]. When the *MAOA-L* variant was detected in 17 out of 46 Maori men, it took very little to attribute to it the warlike attitudes of the entire Maori population and to christen the variant as "warrior gene"[9]. However, successive studies in one of the world populations showing the lowest rates of violent crime, Taiwan, found an even higher percentage of carriers of *MAOA-L*[10]. Far from being a warrior gene, the *MAOA-L* variant is now known to be widespread (of course, in different proportions) in all populations studied so far.

3. DNA evidence on genetic determination of behavior in court

Nobody denies that genes play a crucial role in the development and function of the brain. However, for no gene so far it has been possible to establish a causal relationship with any specific behavioral trait, largely for the complex determination of such traits. Still, there have been multiple attempts to use genetic evidence in court, to claim that a defendant could not be considered fully responsible for her/his actions because such actions were somehow genetically determined. Two rather famous case studies have to do with *MAOA*. In 1991, Stephen Mobley killed John Collins in a pizzeria, in Georgia (USA). His lawyers asked for a genetic test, claiming Mobley could carry a *MAOA* variant predisposing him to violence. The judge stated that no scientific evidence justifies the test, and Mobley was condemned (and ultimately executed in 2005). On the contrary, a judge of the Appeal Court of Trieste innovated world jurisprudence, by granting Abdelmalek Bayout genetic (with T, not with R) extenuating circumstances because, according to the experts, he was "heterozygous carrier of a *MAOA* variant predisposing him to become particularly aggressive under stress situations". Bayout, an Italian citizen, in 2007 had bought a 20cm-long knife, ambushed and stubbed to death Felipe Novoa Perez, who had previously made fun of him. Confronted by the same request as in the Mobley case, the Trieste judge decided instead to ask for an expert opinion. At any rate, the two experts that were chosen, a biochemist and a psychologist, seem to have some problems with genetics, since male cells contain only one X-chromosome, and hence Bayout cannot possibly be heterozygote for any X-linked gene.

Nineteenth century science was deterministic and looked for laws establishing a tight relationship between causes and effects. With exceptions, 21st century genomics recognizes the limitations of our ability to know, and hence is probabilistic[11].

4. Genomic data obtained from patients in clinical studies: is consent really informed?

Despite all hindrances and limitations, the advent of innovative screening and diagnostic tests based on genetic fingerprinting opened the way for radical challenges to the classical concept of evidence-based medicine, shifting towards proactive interventions in the ambitious perspective

[8] H.G. BRUNNER et al., *Abnormal behavior associated with a point mutation in the structural gene for monoamine oxidase A,* in *Science,* 262, 1993, 578-580.

[9] R. LEA, G. CHAMBERS, *Monoamine oxidase, addiction, and the "warrior" gene hypothesis,* in *The New Zealand Medical Journal,* 120, 2007, U2441.

[10] N.J. KOLLA, M. BORTOLATO, *The role of monoamine oxidase A in the neurobiology of aggressive, antisocial, and violent behavior: A tale of mice and men,* in *Progress in Neurobiology,* 194, 2020, 101875.

[11] WILLIAMS R., WIENROTH M., *Social and ethical aspects of forensic genetics: A critical review*, in *Forensic Science Review*, 29, 2017, 145-169.

of a patient-tailored healthcare. Major investments in sequencing technologies and genome-wide association studies (GWAS) allowed the implementation of large-scale genetic datasets with the related health and phenotypic data, enabling geneticists to characterize variants associated with complex traits and common diseases. These approaches may represent a step toward the development of a predictive and individualized, patient-centered medicine. However, ethical and legal constraints underlying these new insights often failed to keep pace with the scientific and technological advancements. While in the next future several millions of individuals are expected to have their genome sequenced, concerns are growing about how consent is obtained, use of the genetic information collected, threats on genetic privacy and risk of discriminations based on DNA signatures. For example, many clinical trials sponsored by pharmaceutical industries envisage the collection of biological material from patients and the sequencing of their genome. These intentions are not always clearly stated in the informed consent that patients sign to enroll in the study, nor is often clearly described the use that the company will make of this information. In other words, large genetic datasets are becoming the property of private companies, that expect to generate profit (not only knowledge) out of them.

Availability of genetic information may entail the risk of discrimination based on identification of particular traits or risk of disease, leading for example to loss of job opportunities or higher insurance rates. Therefore, genetic privacy stands in urgent need for regulation, in particular for genomic data access and sharing but, even more importantly, for the individual's consent to their use. In order to delineate a dedicate framework on personal data at EU level, the General Data Protection Regulation (GDPR) entered into force in May 2018, with the purpose of defining all the details required for personal data sharing at an international level, along with the obligations for individual data usage and processing[12]. However, the advent of the GDPR raised some discussion, in particular with regard to the legitimacy and utility of obtaining such a broad consent.

5. Gene editing

A third genetics revolution has just begun. Products of gene therapy aimed at modifying the gene pool of human cells are already an established and rapidly expanding reality in the clinical practice, with thousands of potential new gene therapies submitted every year to the regulatory agencies[13]. The advent of sophisticated molecular techniques, the most promising called CRISPR-Cas system, makes now possible to easily and precisely "hack" the human genome[14]. There is an urgent need to engage in a public and expert dialogue about the use of these powerful tools. This issue became dramatically clear two years ago, when the Chinese scientist He Jiankui declared to have generated the first gene-edited babies using the CRISPR/Cas9 system, in the attempt of avoiding the vertical transmission of HIV from a seropositive mother. An announcement that outraged the public opinion

[12] L. Marelli, G. Testa, *Scrutinizing the EU general data protection regulation. Science*, 360, 2018, 496-498.

[13] R. Ramezankhani et al., *Two Decades of Global Progress in Authorized Advanced Therapy Medicinal Products: An Emerging Revolution in Therapeutic Strategies,* in *Frontiers in Cell and Developmental Biology*, 8, 2020.

[14] R. Jansen et al., *Identification of genes that are associated with DNA repeats in prokaryotes,* in *Molecular Microbiology,* 43, 2002, 1565-1575; H. Li et al., *Applications of genome editing technology in the targeted therapy of human diseases: mechanisms, advances and prospects,* in *Signal Transduction and Targeted Therapy,* 5, 2020.

worldwide: "He was widely criticized for ignoring important ethical consideration and exposing the girls to unknown risks for an uncertain benefit", as stated in a Nature magazine editorial[15]. One year later, eighteen scientists and ethicists from all over the world called for the adoption of an international moratorium on all clinical uses of heritable genome editing, suggesting a permanent ban on all germline cell gene editing[16].

6. Patenting of nucleotide sequences

Aside from the undeniable value for the present and future of medicine, an issue arising with the ability to re-shape the characteristics and structure of genes is the protection of the scientist's discovery as an intellectual property, i.e. by means of patents, whenever the requirements of novelty, utility and non-obviousness are met. Gene patents are issued to cover the composition of a specific nucleotide sequence and/or the functional or diagnostic employments of derived products. However, it is debated if products that derive from a gene sequence should be considered inventions or discoveries. Numerous ethical and practical concerns arise on how these patents could be exploited, with reference in particular on the potentially detrimental effect on the process of discovery and development of new diagnostics and therapeutics. Although several theories have been proposed to assess the legitimacy of gene patents, there is still a compelling need of clearly defined rules for genetic patentability. Indeed, the practical criteria for granting authorizations of gene patents are quite different among the various countries, and it remains urgent to develop clear and effective guidelines based on international best practices.

7. Conclusions

There is no simple and handy solution for many of the issues raised in this article. The debate is in progress; often, and not unpredictably, scientists, bioethicists, law experts and legislators tend to pay special attention to specific and different aspects of the problems. Needless to say, any lasting solution will have to be respectful of all points of view, finding a balance between the desire to fully exploit the new opportunities offered by science and the protection of individual and collective rights. This is not an easy goal.

[15] D. CYRANOSKI, *Baby gene edits could affect a range of traits,* in *Nature,* 2018.

[16] E.S. LANDER et al., *Adopt a moratorium on heritable genome editing,* in *Nature,* 4, 2019.

Genetic Information: The individual, the family and the Humankind

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1. Introduction

Genetic information has been scrutinized by scholars for years. Lawyers and multiple legal frameworks intend to define and classify it in order to secure its production, storage and use. Most of the legal and soft law instruments, at national, European and international levels, consider genetic information as a special category of heath information to be particularly protected regarding potential misuses. This cautious, even not always justified by all lawyers, can be explained because of the characteristics of genetic information which is making individuals identifiable, which can be shared with family members or which can also be misused for discriminatory purposes. Beyond this individual dimension, genetic information is also characterized by its ontological one, namely the part we are all sharing as members of the Humankind. This difference by nature of genetic information leads to various principles, values and legal frameworks worldwide which have all the same goal of protecting individuals' rights. In this paper, we will argue that genetic information cannot be seen as a unique concept and that further legal and ethical studies are still needed.

2. Genetic information and the individual

Revealed thanks to the realization of a personal genetic test, individual genetic information can either be used for care purposes or for research. In these two areas, the main issue is to ensure trust between patient and health professionals through the enforcement of individual fundamental rights. In this regards, two core principles are usually covered by laws and regulations: the respect for autonomy and the respect for privacy. When produced and used in the care settings, genetic information is considered as standard of care when a genetic disease is suspected (existing clinical evidence in symptomatic or asymptomatic individuals). Autonomy is at the heart of the patient-health professionals (medical care providers, genetic counsellors, nurses) relationship where the goal is to provide patients with accurate scientific data and practices for the benefit of their health. In that sense, autonomy imposes to medical professionals to clearly inform patients about the test to be performed, the risks and benefits, the expected results and the medical treatment if any. These requirements are considered the basis to ensure that patients have clearly understood the goals and the impact of the realization of a genetic test and to give them the possibility to accept or refuse such a test. Then, autonomy results in the capacity of choice, an enlighten choice, to be formalised into an informed consent which is usually required in written. This formal acceptance through a signature, although the usual way to gather consent in medical care is by oral, emphasizes on the importance for health law to ensure patients to be fully aware of a genetic test's consequences for themselves and for their family members. Written informed consent is also required when the genetic test is used for the needs of a research. However, this formal prerequisite is more inherited from ethics research basic principles but can be reinforced when genetic information is about to be interpreted. Research intends to evaluate the benefit-risk balance to allow researchers to validate their scientific hypothesis. In that case, individual genetic

information is different in nature and cannot be mistaken with validated genetic information used in care. Even though patients are involved in research protocol they are no longer considered only as patients but also as research participants which implies the application of other pieces of law. This change of status is one of the challenges to be faced by health professionals which are in charge of clarifying this issue for the patients notably regarding the return of results of research outcomes. Despite the two frameworks are clearly separated in law, genetic test performed in research can reveal information that are already validated in care and which results can be useful for individual's health. This blurring between care and research is, then, interrogating health professionals' duties to inform patients back and to reincorporate them in a routine care. Laws and regulations adopted several mechanisms to try to articulate these positions but communication of results or of incidental findings is still debated. Several complementary issues are still subject to ongoing discussions when it comes to enforce the protection of privacy. As a basis of the medical care and research relationships, confidentiality of medical information is also protected in most of the national laws. Patient must be ensured that the medical, and genetic, information to be revealed during a medical consultation or a research project will be kept secret to third parties. Thus, professional secrecy is considered to be one of the means to ensure trust between health professionals and patients, and is one of the strongest principle for medical information to remain in the hands of the individual. Health law has enforced this principle for a long time and it has been reinforced through the adoption of Regulations regarding data protection. In that sense, the *General Data Protection Regulation* (Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC), has issued a complete (complicated) legal regime for health data and genetic data to be used in the care and in the research settings. GDPR has provided for the first time a definition of genetic information (Article 4 GDPR), but has not drafted a specific legal regime for their processing: they are part of health data (sensitive data) and are falling under this scope. However, Member States still have the possibility to adopt more protective provisions in their national law for the use of genetic data which will probably lead to a jeopardisation of its framing in Europe. Another question remains unclear under the GDPR provisions, regarding family members' access to genetic information.

3. Genetic information and the family

As already mentioned, family members can have interest in being informed of a genetic information which has been diagnosed in the family. Constitutional genetic information is inherited and thus can be transmitted through procreation or can exist in several family members. To date, legal frameworks usually tend to protect individual fundamental rights and have less identified family members as potential rights' owner. The place and rights of family members regarding the access to an existing genetic information is differently addressed in laws, countries balance between no regulation and detailed regulation to solve the dilemma between ensuring the respect of the professional secrecy and ensuring the respect of the duty of family members to be informed of an information which could impact their health. In the countries where the laws are unclear or are not covering this issue, professionals have adopted a case by case approach based

on their deontological rules and ethical principles. In that cases, the legal interpretations of professionals' duties and family members' rights have been provided by judges and courts on which genetic information can or cannot be disclosed to family members. In the countries where the law has precisely tackled this question, usually a procedure is in place in order to allow the transmission of genetic information to family members. In line with the spirit of the Council of Europe *Additional Protocol to the Convention on Human Rights and Biomedicine*, concerning Genetic Testing for Health Purposes (2008), all necessary information about the consequences of the result for the family members should be provided to the patient prior the realization of a genetic test. For example, according to the French law, patient is in charge of communicating with family members (identified with the professionals) thanks to a document prepared by the health professionals. Where patients do not want to communicate with the identified family members, the procedure allows medical doctors to communicate with family members without being prosecuted for breach of professional secrecy. In the latter case, professionals are, thus, more protected regarding the release of genetic information to family members than in countries where no regulation exist. Although several legal positions have been adopted in Europe and worldwide, this duty to inform family members is still debated. This reveals the need to acknowledge that family members can claim rights to access an information which is, by essence, confidential and is part of the individual privacy. Family members are recognized to be rights' owners and should claim a loss of chance of being diagnosed or prevented of a genetic disease. This legal responsibility issue (of the index subject, of the professional) needs to be further studied.

4. Genetic information and the Humankind

As science advances, the ontological part of genetic information is also now challenged. Humankind has been recognized for a long time as subject of rights through the qualification of "crime against the humankind" (first mention in the International Court of Nuremberg Charter, 1946), with the aim of preventing part of populations of being subjects to serious acts committed as part of a widespread or systematic attack directed against any civilian population, with knowledge of the attack' (article 7 of the Rome Statute of the International Criminal Court, 1998). In the field of human genetics, Humankind was first mentioned by UNESCO in the *Universal Declaration on the Human Genome and Human Rights* (1997) where in its article 1 it recognizes "The human genome underlies the fundamental unity of all members of the human family, as well as the recognition of their inherent dignity and diversity. In a symbolic sense, it is the heritage of humanity". However, this Declaration is less providing full rights to the Humankind, than enforcing individual rights with regards to biology and medicine progresses. Only article 11 refers to practices that are considered to be contrary to human dignity such as reproductive cloning. At the time of the adoption of the Declaration, animal cloning was just performed and one the goal of this instrument was to ensure the practice will not be developed in humans in order to protect the unity of the Humankind and most of the national laws have adopted this principle. In addition, several rules were adopted in order to promote research activities that had to be respectful of fundamental rights. The debate on how to protect humankind came back into the bioethics community when CRISPR-Cas9 genome editing technology was

developed and published in 2012[1] Thanks to this technology, it is now possible to modify the genome of embryos and, thus, to modify genetic information that could be transmitted over generations. Several declarations, statements, positions were adopted worldwide in order to assess and frame the potential consequences of this innovation at individual and Humankind levels. However, in these diverse documents, Humankind is only referred to in order to protect its unity whereas individual rights and those to allow biomedical research are largely tackled. Thus, considering this approach it could be useful for lawyers to provide legal clarifications on what could be needed to map new rights for the Humankind. One possibility could be to refer to the project of *Universal Declaration of Humankind rights* proposed in 2015[2]. According to this project,[3] initially devoted to environment matters, Humankind and future generations could be subject of rights (e.i. article VII - *Humankind has the right to protection of its common, natural, cultural, tangible and intangible heritage*) and we, the present generation, have duties towards the Humankind (e.i. article XI - *The present generations have a duty to ensure respect for the rights of humankind as well as of all living species*). The added value of this proposal is to provide a new framework to think together individual and Humankind rights (article XI - *The rights of mankind as a whole and human rights are indivisible and applicable to all future generations*). Another example could also be taken from the French legislation on Bioethics. Thanks to the first laws adopted in 1994, French legislators drafted a new crime in the penal code regarding modification of the ontological genome alongside the existing Humankind crime: the crime against the human species. This new category is characterized by the intention to misuse biological techniques in order to clone or to organize eugenics practices. This offense is one of the most punishable in the French penal system (30 years' imprisonment and 7,500,000 euros of fine). Thus, these frameworks could serve as a basis to more elaborate on potential Humankind rights in the field of genetic information and to adopt a more global vision of the future needs in this field.

5. Conclusion

To conclude, genetic information is a multi-faceted notion which legal regimes should be crossed with its multiple usages in the health system (healthcare, research, public health) and outside (Direct-to-consumer genetic testing). We can observe that the current evolutions of genetic information legal frameworks, tend to incorporate several innovations either scientific (constitutional genetics, somatic genetics) or societal (access to genetic information for the whole population). These are constituting challenges, as it is usually difficult to align regulations and scientific advances, where an enlargement of owners' rights from individuals to family members and may be to the Humankind can be part the pathways towards acceptable, responsible and agreed solutions.

[1] M. JINEK, K. CHYLINSKI, I. FONFARA, M. HAUER, J. A. DOUDNA, E. CHARPENTIER, *A Programmable Dual-RNA-Guided DNA Endonuclease in Adaptive Bacterial Immunity*, in *Science*, 2012; DOI: 10.1126/science.1225829

[2] Available at: http://droitshumanite.fr/?lang=en.

[3] C. DUPRAS, Y. JOLY, E. RIAL-SEBBAG, *Human rights in the postgenomic era: Challenges and opportunities arising with epigenetics,* in *Social Science Information*, 59, 1, 2020, 12-34

Basta con il biolaw: What about knowledge and trust?

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Lasciate ogne speranza, voi ch'intrate
Abandon all hope, ye who enter here

Canto I, Inferno, Dante Alighieri

1. Introduction*

One could be forgiven for surveying the current landscape of norms that regulate genetic data and having the first line of Dante's *Inferno* in the back of one's mind. The terrain is complex and is full of unsightly scenes. And unlike the mellifluous *terza rima* of the magnum opus, the going is tough and there is no guarantee that one will emerge to see again the stars.

The COVID-19 pandemic exists within this universe of norms. The crisis has brought into sharp relief the heterogeneous nature of the public interest rationale in our normative systems. The public interest is what grounds (justifiably) draconian measures such as enforced isolation, mandated wearing of masks in public, and other such incidents that are now part of our new normal. Yet, social life will only again be recognizable through advances in basic and translational science.

COVID-19 host genetics research, whereby the genome of the person infected with COVID-19 is sequenced and analyzed, holds much potential to further our understanding of the variability in response to SARS-CoV-2 and, in turn, to improve clinical care. Following a spring 2020 call for international COVID-19 data sharing and solidarity from the World Health Organization, Wellcome, the European Commission, and others, where is the much-needed international data?

Drawing from recent biolaw events, both pandemic and non-pandemic, we will highlight some of the traps of genetic exceptionalism (1) and knots of data protection (2) that genomics currently faces. We then turn to a creation of a ladder of knowledge and trust to possibly deliver us from the status quo (3).

2. Traps of genetic exceptionalism

One key, ongoing issue as regards the ethico-legal dimensions of genetic data relates to genetic exceptionalism. At its core, genetic exceptionalism posits that genetic data merits different treatment in law and ethics by the very fact that the information is genetic. It has been called into question by others. Genetic exceptionalism is a powerful social, political, legal, and ethical idea that can fuel the propagation of norms. By way of illustration, the Supreme Court of Canada recently upheld the constitutionality of the federal *Genetic Non-Discrimination Act* in part because the "potential for genetic test results to reveal highly personal information about the individual tested and their relatives is immense"[1] and

* *The authors thank Mark Rothstein for his comments on an earlier draft. The authors are also grateful for the support of the Canada Research Chair in Law and Medicine.*

[1] *Reference re Genetic Non-Discrimination Act* 2020 SCC 17 per Abella, Karakatsanis, and Martin JJ at para 88.

because genetic information is "uniquely elemental to identity, and uniquely vulnerable to abuse".[2] The intensity of language that the members of the Court employ raises eyebrows and underscores genetic exceptionalism.

Indeed, nowhere is genetic exceptionalism more evident than in the past 25 years of legislation prohibiting "genetic" discrimination. Such legislation seeks to protect access to health, life, and disability insurance and prevent discrimination by employers. Yet, the only evidence we have thus far of genetic discrimination having a material effect on people's lives is in the realm of life insurance. Individuals with a family history of genetic illnesses will forego essential presymptomatic testing for fear of being denied life insurance.[3] The fear that genetic discrimination inspires may lead to overbroad laws that essentialize genetic information and which do not serve their purported policy objectives.

Singling out genetics under law can actually create additional problems of justice and fairness as discrimination based on physical and mental disabilities has already been long prohibited under human rights law and could well be an avenue for any genetically based injustice. Genetic exceptionalism may further exacerbate the social stigma and perceived "abnormality" of genetic conditions as distinct from other medical conditions. Singling out genetics over other health information may also give a false sense of security. With the -omics revolution, other unique, probabilistic insights can be drawn about individuals, such as with polygenic risk scores, and on which important individual-level decisions may be made. Current approaches to genetic discrimination tend not to have sufficient flexibility to protect against multi-faceted informational and discriminatory harms.

This biolaw trend of singling out genetic data has had a spillover effect beyond the domain of genetic discrimination. Explicit participant consent is typically required to examine the role of genetic factors in research. For prospective studies, this is not an issue. Where, however, important retrospective data analysis is carried out in the public interest, issues may arise. For example, host genetic data from COVID-19 patients is missing from the early months of the first wave of the pandemic. Samples and data collected prior to the creation and ethics approval of multi-site research endeavours had no possibility of prospective participant consent. Although still too early to take stock of the effects the inaccessibility of this data has caused, a postmortem is in order when we are on the other side of the pandemic.

3. Knots of data protection

Related to genetic exceptionalism, the normative claims of data protection over genetic and health-related data have the scientific community in a bind. We have plenty of biolaws, but few, if any, aim to *promote* international collaboration in a way that strikes a proportionate balance between the fundamental data protection and privacy interests of individuals and the collective interest of humanity in the fruits of scientific research. Gone is the age where scientific advancement is the brainchild of a handful of individuals whose data scarcely left their lab notebooks. Instead, insights are drawn through intense, international collaboration where the broad sharing of data is needed.

[2] *Ibid.* per Abella, Karakatsanis, and Martin JJ at para 92.

[3] M. ROTHSTEIN, *Can Genetic Nondiscrimination Laws Save Lives?*, in *Hastings Center Report*, 51, 1, 2021.

Instead of biolaw that supports collaboration, we witness the imposition of cumbersome legal processes on data sharing between institutions and countries, due to the already existing data silos and legal drawbridges. Data linkages need to be robust and secure, but also efficient and low-cost. Genetic and familial medical records and administrative health data should be further linked. While after a decade, national resources such as biobanks have succeeded in lowering some of the legal drawbridges, the same is not true for the transfer of medical and genetic data across jurisdictions.

Consider the European Data Protection Board (EDPB)'s statement on COVID-19 research. The EDPB merely states that international cooperation is "probably" required and that international data transfers "may" be implied.[4] (To say nothing of the complexities genetic and health-related research faces for international transfers following the *Schrems II* decision.) The EDPB's guidelines "lack both any sense of urgency and any consideration of the public good, and fail to take into account other fundamental rights, societal interests, and scientific considerations".[5] By and large, current approaches to the *General Data Protection Regulation* have lost sight of its recitals, which are meant to guide its interpretation and emphasize the need to strike a proportionate balance of fundamental rights and to promote the welfare of citizens.

4. A ladder out: prioritizing knowledge and trust

Policy making requires a strong social nexus. Generalized social sensitivity to genetic data can give rise to a "prohibition reflex", whereby with certain advances, the public swiftly demand a law against "it".[6] Yet, despite these pressures, there is little empirical data regarding the public's attitudes about the sharing of genetic data and of their genetic data literacy. A first-of-its-kind global survey of 36,268 individuals across 22 countries (and in 15 languages) indicated that those individuals who had greater knowledge of genetics and trusted the users asking to use their genetic data were more likely to be in favour of donating data.[7] This suggests that knowledge and trust are central to delivering on the promise of genetics. Both are fiercely difficult to establish. A simple prohibition does not serve the former and only serves the latter when the prohibition is tailored to preventing behaviour that is repugnant to social expectations.

Promoting knowledge calls for a reasoned look at genetic data to take stock of its features and implications. *Any* data type has its own distinguishing characteristics. Indeed, a taxonomy of data would not be possible without recognizing such characteristics. Rather than saying *simpliciter* that genetic data is different, we can look at the ways in which it is functionally different. Yes, genetic data can give us probabilistic insights about one's future health status. But, so does one's postal code (as a proxy for both

[4] European Data Protection Board, *Guidelines 03/2020 on the processing of data concerning health for the purpose of scientific research in the context of the COVID-19 outbreak*.

[5] J. Bovenberg et al., *How to fix the GDPR's frustration of global biomedical research*, in *Science*, 370/6512, 2020, 41.

[6] B.M. Knoppers, *Scientific Breakthroughs: The Prohibition Reflex (From IVF to AI)*, 2019 Friesen Lecture at the University of Ottawa, available at https://www.youtube.com/watch?v=TnA6f4Jr2FI. See also B.M. Knoppers, *Does policy grow on trees?*, in *BMC Medical Ethics*, 15, 87, 2014.

[7] A. Middleton et al., *Global Public Perceptions of Genomic Data Sharing: What Shapes the Willingness to Donate DNA and Health Data?*, in *The American Journal of Human Genetics*, 107, 2020, 743-752.

environmental and socio-economic factors). Indeed, even conditions with significant familial aggregation, such as type-2 diabetes and obesity, are increasingly understood as the result of complex relationships between and among genetic, epigenetic, and environmental factors.

Further reflecting on the relational aspects of genetic data, familial implications emphasize the importance of examining the ambit of the (expanding) duty of care that clinicians owe not only to patients but also to family members. Potential ways to mediate among competing priorities such as the right not to know and the duty to warn must also be considered and translated into appropriate contexts through tools such as familial consent clauses. Perhaps, however, such work is better contextualized within a broader debate that patients need to be in dialogue with their genetic relatives and relevant healthcare professionals regarding the implications of any diagnosis or risk factor for others.

Increasing scientific literary, and genetic literacy in particular, requires broad coordination between the State and civil society in providing both accurate, accessible information and the creation of education opportunities for children, adolescents, and adults alike. Indeed, awakening the human right to science implicates better scientific education. Beyond prohibitions, trust requires that what actually happens with genetic data accords with the expectations of interested individuals and groups. One hopes that, in the *longue durée*, the dialectic between knowledge and trust will create evidence-based policy that supports international collaboration.

5. Conclusion

Genetic testing is no longer only for individuals with rare diseases, nor for research purposes alone. With the development of clinical genomics services, the importance and relevance of genetic information for medical diagnoses and care is greater than ever. Stewardship of this area of biomedicine calls for policy that supports knowledge and trust of actual and future patients and citizens alike.

We have come a long way from the 1996 Bermuda Principles, which mandated the rapid, open release of genetic sequences for the benefit of humanity. In the intervening years, we have become an information society with acute concerns about unscrupulous uses of personal data. Concerns about informational harms, discrimination and stigmatization, combined with notions of genetic exceptionalism, have largely caused policy discussions to lose sight of the need to secure benefits alongside reducing the likelihood of harms.

Any biolaw, then, must have this concern of proportionately balancing risks and benefits at its heart. Autonomy and privacy must be furthered alongside strong, open science. Long ago, scholars identified that genetic data implicated the interests of multiple groups such as the proband, their genetic family, healthcare professionals, researchers, the State, insurance companies, and others. The proband's prerogatives will be the starting point, but they alone cannot determine the course of policy making. Rather, any sensible normative framework must balance these varied interests in a way that is sensitive to context and to a society's core values and principles.

The limitations of (statutory) law should also be recognized. Positions may crystallize in a way that is increasingly in tension with contemporary scientific knowledge. Do not let overly simplistic accounts of genetic exceptionalism fool you. Complexity must be embraced in conjunction with normative flexibility in the face of such a rapidly progressing domain.

We encourage lawmakers and policymakers alike to seize upon the entire normative toolkit

at their disposal: policies, standards, memoranda of understanding, regulations, international declarations, codes of ethics, and, if appropriate, primary legislation (statutes). These norms should not claim the entire decisional space. There must be room for transparent, professional discretion combined with effective mechanisms of accountability. If anything, the COVID-19 pandemic highlights the urgency to ensure that clear pathways exist for the invocation of the public interest by trusted experts who are accountable to the public.

ISSN 2284-4503

Overcoming legal obstacles to international direct-to-participant genomic research*

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The relationship between genetics/genomics and the law has been varied, complicated, and continually evolving. In some areas, such as eugenics-inspired sterilization laws, it has been disastrous. In other areas, such as newborn screening and DNA forensics, legislation has been generally beneficial. In research and clinical applications, the record has been mixed, containing legislative successes tempered by occasional failures that serve as cautionary tales. Such is the case with laws applicable to international genomic research. This article reviews legal regulation of international direct-to-participant (DTP) genomic research as a case study of the challenges of combining science, law, ethics, and other issues.

Among the most important recent trends in genomic research are international collaborations and DTP recruitment of research participants. The combination of these two elements is essential for research on rare disorders because there may not be enough affected individuals in any country to study and genomic heterogeneity is often a key element of the research strategy. The use of DTP recruitment using the internet, especially when endorsed and supported by patient advocacy groups, has received approval from institutional review boards (IRBs) and research ethics committees (RECs) for national studies, and it has proven to be effective in genomic research compiling and analyzing biospecimens. It is usually far more challenging, however, to obtain a positive research ethics review for international studies because the scope is greater and the legal systems in numerous countries vary considerably and may be difficult to discern. Consequently, the prospect of needing separate approval of a research protocol in every country where only a few individuals may participate will make international recruitment impractical and thwart genomic research on rare disorders.

This article draws lessons from a 31-country study of international DTP genomic research funded by the National Institutes of Health of the United States and published in 2019.[1] The study deals with the conceptual and practical challenges of promoting international genomic research while ensuring compliance with the letter and spirit of international laws and local norms governing research ethics. It also serves as a case study for the larger issues of reconciling legal principles around the world to enable genomic research.

Assessing the Laws in Numerous Countries

Researchers undertaking genomic research on rare disorders face many legal challenges in expanding their efforts to multiple countries. To begin with, there is no easy way to learn what, if any, laws around the world apply to foreign researchers soliciting individuals to participate in

* *The research described in this article was supported by grant No. 5R01HGO09914, Regulation of International Direct-to-Participant Genomic Research, from the National Human Genome Research Institute of the U.S. National Institutes of Health, Mark A. Rothstein and Bartha Maria Knoppers, Principal Investigators. The complete set of 34 papers drafted for this study are contained in the following symposium:* Regulation of International Direct-to-Participant Genomic Research, in Journal of Law, Medicine & Ethics, 47, 4, 2019.

[1] M.H. ZAWATI, ED., *Country Reports*, in *Journal of Law, Medicine & Ethics* 47, 4, 2019, 582-704.

ISSN 2284-4503

genomic research or facilitating the submission of biological specimens and health information. Some compilations of international research laws and regulations are published periodically,[2] but most of the information sources rarely cover emerging issues, such as DTP recruitment, and published information is soon out of date.

Next, there are language and translation issues. For example, a law might provide that "consent" is required, but consent has numerous meanings, such as informed consent, written consent, broad consent, and presumed consent. Therefore, it is still necessary to identify and retain legal experts from every country in which research is proposed to explain the precise meaning of a law or regulation. The difficulty, time, and expense of such inquiries in numerous countries imperils international research.

In our study of DTP genomic research, we learned that very few countries have enacted laws directly on point, which was not surprising because of the recency of DTP online recruitment. Therefore, we had to extrapolate from existing laws (e.g., direct-to-consumer genetic testing laws, human subjects research laws, and genetic privacy laws), to infer the likely legal position of each country on DTP genomic research. Furthermore, we needed to identify "soft" law in the form of regulations, norms, and cultural considerations so that proposed research would align with a broad array of values in each country. Fortunately for our study, we were able to assemble an incomparable group of 45 experts to prepare the country reports that led to the study's conclusions and recommendations.

Equivalency of Common Principles

Early on, it became clear that an international treaty or a series of bilateral agreements would be infeasible. Even assuming there was international support for this approach (a grandiose assumption), it would likely take years to negotiate and implement such an agreement and, in the interim, potentially valuable genomic research would be significantly delayed. A more expeditious and practical strategy would be to build on existing, country-specific legislation using a two-step process of identifying a generally concordant legislative or regulatory framework and then devising a method for its multilateral application. Every country we studied had already enacted laws regulating research with human subjects based on a similar set of underlying principles. The United Nations Educational, Scientific and Cultural Organization (UNESCO), in its Universal Declaration of Bioethics and Human Rights, specifies the broad criteria for ethics review, including informed consent, privacy/confidentiality, benefit/risk ratio, return of results, protection of the interests of vulnerable persons/communities, and research integrity and safety.[3] Another key, non-governmental document is the Global Alliance for Genomics and Health (GA4GH) Ethics Review Recognition Policy. Designed to regularize international genomic research review, it is based on ethics review policies of 39 countries. The foundational principles are: respect individuals, families, and communities; advance research and scientific knowledge; promote health, wellbeing, and the fair distribution of benefits; and foster trust, integrity, and

[2] Office for Human Research Protections, U.S. Department of Health and Human Services, in *International Compilation of Human Research Standards*, 2020, https://bit.ly/324OSsj (last visited 08/11/2020).

[3] UNESCO, *Universal Declaration of Bioethics and Human Rights*, 2005, https://bit.ly/3g2GH7P (last visited 05/11/2020).

reciprocity.[4] We also reviewed several other sources, including the World Medical Association's Declaration of Helsinki[5] and the Council for International Organizations of Medical Sciences (CIOMS)-World Health Organization (WHO) International Ethical Guidelines for Biomedical Research Involving Human Subjects.[6]

We drew from these important documents and other sources additional principles with international applicability. First is the requirement of establishing an independent, external body to perform research ethics review. In the United States, this is called an institutional review board because review is conducted largely within a single institution. In much of the world these bodies are called research ethics committees. Second is the growing trend to have single-site ethics review for multi-site studies, as multiple ethics reviews tend to cause needless delay without advancing the welfare of research participants. In the United States, as of 2019, single-site or "central" IRB review is required for multi-site studies.[7] Third is recognition that international DTP genomic research is low risk. Participants typically spit into a vial or swab the inside of their cheeks (or merely supply data), and there is no intervention or alteration of their medical care. Fourth is acknowledgment that DTP genomic research, especially for rare disorders, is overwhelmingly supported by affected individuals and their families. Removing needless burdens on researchers, such as requiring research ethics review in every country, advances the autonomy interests of participants and their caregivers.

Based on these principles, my colleagues and I concluded that the most promising, basic approach would be to have international ethics review undertaken by a single entity (i.e., IRB or REC) in the researcher's country. There were "only" two practical questions. First, why would the home countries of the potential research participants agree to defer to an approval by the researcher's ethics review body? Second, how would the researcher's ethics review body know whether the research protocol would be lawful and ethical in the various countries where participants might be enrolled?

Adequacy Determinations

The next step was to envision how deferral agreements could be reached without reconsidering the issue each time a new research protocol was submitted. We looked to the principle of "adequacy" under the General Data Protection Regulation (GDPR) of the European Union (EU).[8] Article 45 of the GDPR provides that personal data may be exported to a country outside of the EU only if the European Commission (EC) has acknowledged the adequacy of data protection in the recipient country or there are other appropriate safeguards such as contractual provisions or codes of conduct.[9] Adequacy based on the equivalency of another country's laws is determined by reference to the principles noted in the

[4] Global Alliance for Genomics and Health, *Ethics Review Recognition Policy*, 2017, https://bit.ly/3mFIu4f (last visited 05/11/2020).

[5] World Medical Association, *WMA Declaration of Helsinki – Ethical Principles for Medical Research Involving Human Subjects,* 2013, https://bit.ly/3a4knHc (last visited 05/11/2020).

[6] Council for International Organizations of Medical Sciences (CIOMS) and World Health Organization*, International Ethical Guidelines for Health-Related Research Involving Humans*, 2016, https://bit.ly/3t64x6p (last visited 05/11/2020).

[7] 45 C.F.R. § 46.114, b, 1.

[8] Regulation (EU) 2016/679 of the European Parliament and of the Council.

[9] J. WAGNER, *The Transfer of Personal data to Third Countries under the GDPR: When Does a Recipient Country Provide an Adequate Level of Protection?,* in *International Data Privacy Law,* 8, 4, 2018, 318-337.

GDPR that must be satisfied for an adequacy determination by the EU. Two illustrative principles are:

"7. The foreign country's legislation should include basic data protection concepts and remain consistent with the general principles enshrined in the GDPR;

8. Data must be processed in a lawful, fair, and legitimate manner while being set out in a sufficiently clear manner".[10]

Using these criteria, Argentina, Canada, Japan, New Zealand, Switzerland, and other countries have been deemed adequate by the EC for transfer of data from EU countries.[11] My colleagues and I concluded that an analogous regime could be successful for approving international research, especially as applied to DTP genomic research.

Recommendations

We presented our conclusions, rationales, and supporting documents in an article containing the following recommendations.

1. International DTP genomic research approved by an ethics review body in the researcher's country should be deemed approved in the participant's country if the ethics review in the researcher's country has been determined to be adequate by the participant's country.
2. To facilitate international DTP research and to inform potential researchers and participants, a list of countries whose ethics review is deemed adequate should be posted on the website of the regulatory authority responsible for the ethical conduct of research with human participants, such as the OHRP in the United States. Compilations of these country-developed adequacy determinations by international organizations would facilitate international reviews.
3. Ethics review bodies evaluating proposals for international DTP genomic research should consider whether the countries from which participants will be enrolled accept single-site ethics review in the researcher's home country.
4. Ethics review bodies reviewing proposals for international DTP research submitted by researchers in their home country should evaluate whether the researchers have given due regard to cultural considerations in the countries from which participants will be enrolled.
5. Regulatory authorities responsible for the ethical conduct of research with human participants should inform ethics review bodies under their jurisdiction of the approval criteria for international DTP genomic research.
6. Additional research is needed to assess the socio-cultural implications of international DTP genomic research in various population subgroups, including minority and indigenous populations.[12]

Conclusion

The symposium issue of a leading journal containing the country reports and recommendations was published at the end of 2019. We organized a series of presentations for researchers, research regulators, patient advocates, and

[10] ARTICLE29 Newsroom – Working Document on Adequacy Referential (Wp254rev.01) – European Commission, https://bit.ly/3wMSA7S (last visited 08/11/2020).

[11] Adequacy Decisions, European Commission, https://bit.ly/2OEommg (last visited 08/11/2020).

[12] M.A. ROTHSTEIN et al., *Legal and Ethical Challenges of International Direct-to-Participant Genomic Research: Conclusions and Recommendations, in Journal of Law, Medicine & Ethics*, 47, 4, 2019, 705-731, 723-724.

international governments for 2020. Unfortunately, the coronavirus pandemic precluded holding these events, which we hope to reschedule for 2021. In any event, I believe our experience in attempting to facilitate international genomic research in a manner consistent with the laws of numerous countries illustrates the range of conceptual and practical issues to be addressed by laws dealing with genetic technologies.

ISSN 2284-4503

Gene editing: Do we need a universal approach?*

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Only a few years ago, a new scientific discovery shocked not only the scientific community, but also society as a whole: gene editing using the CRISPR Cas9 technique. Similarly to what happened when it was reported that a mammal was obtained by means of an ingenious technique (transferring the nucleus of a somatic cell into a previously enucleated egg cell).

In contrast to cloning, whose spectrum of possibilities focused on reproduction and shortly afterwards on research to treat certain pathologies (the misleadingly misnamed "therapeutic cloning"), gene editing is proving much more promising than cloning was in the past. Gene editing is opening up an almost inexhaustible range of applications, not only in direct relation to human biology (the prevention of hereditary diseases, their treatment and possibly even procedures to improve or enhance already born human beings and their offspring), but also with respect to other non-human living beings. All agree that it is a relatively simple, cheap and efficient technique, although these are under discussion.

As in the past, scientists (and the institutions and companies behind the funding of the research) are pressing for applying this technique to human beings, both in the somatic and germ line. For their part, ethicists, lawyers and policy-makers face similar dilemmas to those of the past, based on different techniques: should gene editing be allowed in the human germ line, or should it be rejected altogether? What medical, moral and legal criteria should we keep in mind to distinguish and assess the permissibility of the use of gene editing for preventive, therapeutic or enhancement purposes?

The knowledge and experience gained in the more than twenty years since a similar dilemma arose with human reproductive cloning, both scientifically and in the field of normative sciences, mainly Bioethics and Law, has taught us some lessons; whether we have been able to learn them is another matter. It would suffice to recall reproductive cloning, at the time full of emergencies, as it could satisfy the supposedly pressing need of thousands of couples to have children. Finally, it was stopped when it was found that this technique was not so easy to apply to humans, unlike other mammals (without going into the significant side effects it had on them), that other reproductive techniques already known at the time were more efficient and safer. Cloning for research purposes was also abandoned because of its own technical difficulties; something similar happened with research with totipotent human embryonic cells.

Firstly, these issues have shown that extreme positions should be avoided, as they lead to closing the doors to further reflection and social dialogue. New information and new approaches are always emerging that can lead to a change of perspective, albeit in a moderate way.

Secondly, in this form of gene editing as in other genetic engineering techniques on germ-line and

* *This work is carried out within the framework of the Basque Government funding for Research Groups in the Basque University System (IT 1066-16).*

in reproductive cloning, the main problem that is (or was) to be solved is not to save the life of a patient, even in the foetal stage, nor to improve his or her health in any significant way. The aim of this technique is to ensure that a future child, not yet even conceived, will be engendered and born free of the diseases that his or her parents are carriers of. In other words, this is not a vital or urgent matter for specific individuals. Moreover, at the moment it does not seem foreseeable that the CRISPR Cas9 technique will be available in the short term to be applied to humans, at least in the germ line.

These facts should lead us to conduct ourselves with caution, reflection and in a measured manner.

Thirdly, as in other sectors of human productivity, the globalisation of scientific research has developed, with high-level scientific projects often being carried out with the concurrence of a plurality of research groups located in centres of excellence throughout the world. Consequently, what were known in the 1990s as "genetic paradises" are now less of a concern from the point of view of the regulatory framework. This term was intended to describe the risk for states with specific restrictive regulatory frameworks and even for international organisations (think of the important legal instruments adopted by UNESCO, the WHO and the Council of Europe, for example) that other countries without legislation, not characterised by their leadership in the fields of scientific research in human biotechnology, would welcome foreign scientists and companies to their territory, thus causing companies to translocate to these countries.

The current concern is to find regulatory frameworks or other procedures for monitoring and control of these activities that are generally and universally accepted and shared. It is also a matter of concern that some states that are particularly prominent and dominant in international politics refuse to apply international agreements to their respective research collectives, in view of the great economic importance that many of these activities promise in their industrialisation and commercialisation phase. We have examples of great powers with dictatorial regimes (e.g. P.R. China), or that do dubiously democratic (Russia) or ultra-liberal (occasionally the USA) practices that do not always respect agreements of various kinds or recommendations to limit certain activities until minimum points of international consensus are found.

Finally, society claims that the moral assessment of these matters and the establishment of regulatory frameworks, since they can have a radical impact on the essence of the human being as a moral entity and as holder of fundamental rights, cannot remain confined to the circle of reflections and decisions of researchers, health professionals or their respective scientific societies. The whole of society is concerned, both individuals and the political (states), cultural and other collectivities in which human beings are integrated.

There are issues under discussion which are of the utmost relevance, but for which cultural, ideological, religious and social diversity does not allow for global agreements to be reached without great difficulty. Thus, the meaning of human life, human beings' belonging to their species and the safeguarding of this in what may be their essence, the moral and legal aspects arising from these recognitions, such as the moral status of human life, the right to life and to physical and moral integrity, the rights related to the human species, our responsibilities towards future generations, the specification of the scope of human dignity in the context of the modification of one's own and individual genetic endowment as part of one's biology, etc. For the time being, it

seems that these issues should be left in the reserve of the debate, which will have to be long and will have to be broadened as progress is made in reaching important, but less ambitious, consensuses.

Given this current scenario, as opposed to the maximalisms of the past, there are controversies for which it seems easier to reach consensus. For the time being, it seems advisable that points of discussion focus on specific issues, on which agreement is easier to reach in the early stages of the global dialogue. For example, the safety of the gene-editing technique, its reliability, the occurrence of anticipated or unanticipated side effects, is of general concern, as it has been shown that there are aspects related to it that are still far from being resolved: the side effects that may result from its specific application in human beings (in their reproductive cells, in the zygote and in the early embryo). This is a crucial issue at the moment, although there are always those who argue that it is only a question of time, of researchers finding a safe way to prevent major risks for the new being. It is also claimed that reliability and efficiency is just a matter of time, of continuing research. This is probably the case, but it does not exempt us from paying due attention to it and taking whatever measures are necessary, even if they are provisional and revisable, to prevent risks and ensure a reasonable level of efficiency, so as to prevent guinea pig behaviours.

Consequently, proposals for universal dialogue are increasingly being made by numerous international organisations, as well as international or supranational conferences, expert groups and ethics committees. The Council of Europe's Bioethics Committee (DH BIO) and the European Commission's European Group on Ethics in Science and New Technologies (EGE) (Opinion on the Ethics of Genome Editing), for example, are committed to this approach. This is notwithstanding the fact that the Council of Europe Convention on Human Rights and Biomedicine (Oviedo Convention) states that "an intervention seeking to modify the human genome may only be undertaken for preventive, diagnostic or therapeutic purposes and only if its aim is not to introduce any modification in the genome of any descendants" (art. 13). UNESCO has also openly declared its rejection of this type of intervention ("[...] in particular regarding the identification of practices that could be contrary to human dignity, such as germ-line interventions", art. 24).

The wording of Article 13 of the Oviedo Convention has generated intense debate over the last few years, and has given rise to various proposals, including: maintaining it in its current wording; reaching a consensus on a more open interpretation of this provision, with the risk of distorting its current legal meaning, whether one likes it or not; and amending this article (e.g. by means of an Additional Protocol to the Convention), in such a way as to allow certain interventions on the human germ line, for example for preventive or therapeutic purposes against serious or very serious diseases, subject to the necessary controls, even if it means modifying the genome of the unborn human being.

It appears to me that it is still premature to examine the need to revisit in short-term the permanence of article 13 in its current wording, given that we still do not know very well what the development of this technique applied to the germ line may be, in particular its reliability and efficiency, to the point that it may one day be applied with sufficient margins of success and safety, without going into the more fundamental issues mentioned above. This is a clear example of the fact that Law must follow Science, there is no need to rush, notwithstanding to maintain an open dialogue on this issue.

This perception of how to deliberate and make decisions or, where appropriate, propose recommendations to authorities and legislators has been understood and practised for several decades through standing committees and ad hoc working groups, ensuring that their composition is multidisciplinary, independent and ideologically and culturally pluralistic and inclusive. However, the application of gene editing in humans (and other living beings) is now regarded as a global issue, which requires global governance, although achieving this goal seems still far from being achievable. Various proposals point towards the creation of an independent global committee that for the time being would limit itself to giving recommendations to researchers on what would be acceptable or objectionable from an ethical and scientific point of view, even proposing moratoria.

We already have expert group initiatives, such as the *Expert Advisory Committee on Developing Global Standards for Governance and Oversight of Human Genome Editing*, which was set up in 2018 by the WHO to examine the scientific, ethical, social and legal challenges associated with human genome editing (both somatic and germ cell), which in 2020 published the document *Human Genome Editing: A DRAFT Framework for Governance*, the annex to which raised numerous questions that needed to be answered or resolved in order to implement international governance of gene editing in humans. However, although it is a multidisciplinary committee, it does not seem sufficient to reinforce this requirement and its recognition as a universal independent body.

Another proposal consisting of an international regulatory commission agreed by scientific academies has been considered a premature and problematic approach to governing human germline genome editing; deferring to a single commission to set the agenda for global governance raises troublesome questions of framing and representation.

A worldwide pluralistic and democratic governance calls for a new process of active and sustained dialogue among stakeholders as well as public authorities and society as a whole. However, there are numerous and important problems that would have to be resolved for this hypothetical world committee to be able to work efficiently: defining its structure and composition, its non-binding nature, but its exhortative nature through its recommendations. It would also be necessary to decide to which international body it would be associated or whether it should be detached from any of them, but with the material and moral support of several bodies at the same time (consortium of bodies such as UNESCO, the WHO, the Council of Europe, the European Union, the OAS, the OAU, ASEAN and others like them).

It is true that a formula such as the one proposed here still needs reflection, dialogue and maturation, so that its moral authority to impose its criteria and proposals is recognised.

ISSN 2284-4503

Posthuman dignity and the problem of the body

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This essay examines the anthropology of transhumanist and posthumanist thought, especially its account of human dignity. Particular attention is given to the place of embodiment within its philosophical worldview. It is argued that posthumanism necessarily defines the body as a problem. The body represents a barrier to achieving dignity rather than a site of intrinsic dignity. To become more dignified requires moving beyond the natural inheritance of the body and even embodiment itself. The framing of dignity and embodiment within posthumanist thoughts illuminates, in turn, broader ethical considerations within contemporary biotechnology.

The meaning of transhumanism and posthumanism is subject to debate and confusion. They both concern broadly the ways in which technology can be used to change and improve the human body and even human nature. In an oft cited definition, The World Transhumanist Association has defined transhumanism as "The intellectual and cultural movement that affirms the possibility and desirability of fundamentally improving the human condition through applied reason, especially by developing and making widely available technologies to eliminate aging and to greatly enhance human intellectual, physical, and psychological capacities."[1] This project might encompass a diverse range of ideas, aims, and technological projects ranging from synthetic biology, prosthetics, and genetic engineering, to more fantastical visions of the human future including the radical extension of life and even the realization of digital immortality. The end point of these enhancements is the posthuman condition, which might include an evolutionarily new species or disembodied forms of being. According to this formulation, transhumanism and posthumanism are connected in that transhumanism is the process by which the posthuman is realized. Of primary concern is the objective to move beyond the boundaries of human nature so that human beings can evolve from a biological inheritance to a machine-based future – that is, from homo sapiens to techno sapiens. This line of thought, however fantastical, reveals most clearly the anthropological assumptions that undergird the transhumanist project. It reveals the premises that shape posthumanist understanding of the body's meaning and significance.

Posthuman dignity is a term most often associated with the work of Nick Bostrom, who has argued that making persons more intelligent, more self-controlled, more immune from debilitation or disease, more liberated from the drudgeries of labor, and even free from the grip of death itself is to make them more dignified. Bostrom argues that "it is possible that through enhancement we could become better able to appreciate and secure many forms of dignity that are overlooked or missing under current conditions."[2] Technological enhancement, according to Bostrom, offers the promise of human betterment and a more dignified existence. In this

[1] N. BOSTROM, *Introduction – The Transhumanist FAQ: A General Introduction*, in CALVIN MERCER, DEREK F. MAHER (eds.), *Transhumanism and the Body: The World Religions Speak*, New York, 2014, 1.

[2] N. BOSTROM, *Dignity and Enhancement*, https://www.nickbostrom.com/ethics/dignity-enhancement.pdf (last visited 23/04/2021)

respect, human nature as bequeathed by the evolutionary process is a barrier to dignity. It is the role of humanity to arrogate to itself the work of overcoming the limits of nature in order to become more dignified. As Bostrom puts it, "Transhumanists view human nature as a work-in-progress, a half-baked beginning that we can learn to remold in desirable ways."[3]

As framed by posthumanist thought, dignity is not a given characteristic of human nature. It is not an ontological status bestowed upon persons. Dignity is made and achieved. It emerges through a process, indeed an ongoing and endless process, and can be expanded by improving the human condition. In other words, nature as such has no normativity. The body is the site of pure possibility to be realized. As such, the path to dignification goes through, or perhaps around, the human inheritance and especially the body. Human life is enhanced most fully by overcoming the human. To be liberated from the limits of the human is to become more dignified.

Bostrom's account of dignity contrasts with the so-called bioconserative position. For bioconservatives, nature is normative. There are essential features of our humanness, bound up in the limits of nature, that should be preserved even when technology might permit moving beyond. There is a fixed givenness to the human inheritance that makes moral claims upon us. Changing the essential character of our personhood is an affront to the dignity unique to humans. The limits that define our nature - including the final limitations of suffering and death – are essential aspects of what it means to be human. Their presence within the scope of human life provides occasions of moral reflection about what it means to live this human life with dignity. It is dangerous and hubristic to reject them.

The posthuman account of dignity informs its view of the body. The body is not itself a bearer of dignity and, in fact, might better be viewed as an obstacle. It is the site of vulnerability and suffering. To enhance the body is to diminish that which stains the human condition, and to move beyond the body is to overcome the risks endemic to life. To become less reliant on our bodies – to become, in this respect, less human – is to become more dignified. In a paradoxical way therefore, the achievement of ultimate mastery over human nature requires eviscerating the naturalness of the body.

It might be the case that even in an imagined technological future, persons will remain tethered in some ways to bodies and other forms of physicality. Yet, even if the body cannot be fully overcome then it should at least be mastered, remade, and improved. The human experience of embodiment is already undergoing a revolution. The line between human and cyborg is blurred. Sex dolls, social media, and virtual reality all point towards new ways of being human and of experiencing embodiment – and disembodiment. What it means to be an embodied creature is more complicated and pluralized than ever.

This emphasis on the malleability of the body explains the central role that transgenderism holds within the transhuman movement. Gender, like the body itself, is merely one expression of form. It is something that can be chosen, refashioned, and imposed upon the physical body. The capacity to remake one's biological sex reveals a wider freedom of the self over the body. Transgenderism vests humans with autonomy over gender,

[3] N. BOSTROM, *Transhumaist Values*, F. ADAMS (ed.), *Ethical Issues for the 21st Century*, Charlottesville, 2003, 4.

sexuality, and embodiment and illuminates the basic malleability of our inherited nature. It renders the body an accidental vessel that contains within it something more essential to be liberated. As Martine Rothblatt observes, "A basic transhumanist concept is that a human need not have a flesh body, just as a woman need not have a real vagina".[4] The body is simply a template for achieving authenticity.

There is a gnostic quality to posthuman thought. The decentering of the body is paired with the claim that essential personhood resides in the brain. We are thinking beings not embodied beings. The body thus has no definitive bearing on our identity as persons. The ultimate goal, in fact, is to download the contents of the brain so that thought and identity might continue in the form of digital immortality. Salvation comes not through the resurrection of the body but the resurrection of the mind in a supercomputer. This prioritizing of mind over body is itself hardly novel within the history of thought, but posthumanism vests it with a distinct technological gloss. Posthuman anthropology combines with technological sanguinity to envision a means by which persons might be freed from the limitations and indignities of the body and thereby achieve a higher more dignified form of existence.

This impulse to overcome the body is connected with the posthuman understanding of selfhood. Russell Blackford describes posthumanism as a philosophy of "self-transformation" and "self-overcoming".[5] This use of the language of self is revealing, for in transhumanism the self is both subject and object. The self uses its autonomy to remake itself. Posthumanism is premised on the vision of an open future that has no end in a teleological sense. It is a future defined by autonomy and freedom from the strictures of nature and convention alike. It is not anthropology that limits possibility but only technological feasibility. Along these lines, Andy Clark proposes "that human minds and bodies are essentially open to episodes of deep and transformative restructuring".[6] This invites, in turn, the use of technology to enhance, transform, and ultimately overcome the human. In the end, everything is malleable. Everything is open to manipulation. Everything is the object of will and power.

The irony of posthumanism is that while its looks to the radical possibilities of the human future, it remains solidly wed to the cultural inheritance of modernity. Posthumanism is tethered to modernity's project of domesticating and controlling nature. It represents a radical extension of faith in progress and the possibilities of improving the human condition, along with the attendant belief that such possibilities lie within the human prerogative. It is proposed that through technological mastery the ills of the human condition can be overcome, perhaps to the point of achieving a kind of perfection. Posthumanism extends the Enlightenment pursuit of mastery over nature to mastery over the human. It is, in this respect, the acceleration and fulfillment of modernity's deepest moral ambitions.

In the end, the posthuman project is only secondarily a technological endeavor. It is in the first instance a moral project that seeks to give

[4] M. ROTHBLATT, *Mind is Deeper Than Matter: Transgenderism, Transhumanism, and the Freedom of Form*, in M. MORE, N. VITA-MORE (eds.), *The Transhumanist Reader*, West Sussex, 2013, 318.

[5] R. BLACKFORD, *The Great Transition: Ideas and Anxieties*, in M. MORE, N. VITA-MORE (eds.), *The Transhumanist Reader*, West Sussex, 2013, 422.

[6] A. CLARK, *Re-Inventing Ourselves: The Plasticity of Embodiment, Sensing, and Mind*, in M. MORE, N. VITA-MORE (eds.), *The Transhumanist Reader*, West Sussex, 2013, 11.

expression to a normative account of dignity, happiness, and human flourishing. Posthumanism offers the purest encapsulation of "expressive individualism," a term coined by the American sociologist Robert Bellah and recently appropriated by Carter Snead in his work on public bioethics. Snead summarizes the anthropology of expressive individualism as follows: "In its pristine form, expressive individualism takes the individual, atomized self to be the fundamental unit of human reality."[7] The expressive individualistic self is defined by an emphasis on choice and authenticity, autonomy as opposed to relationality, mind over body, and the evasion of vulnerability. This theoretical framework might well be applied towards evaluating issues raised by transhumanism and posthumanism. Indeed, examining the anthropology of transhumanism on these terms illuminates the outworking of deep entrenched cultural assumptions that might remain more occluded in other biotechnological contexts.

In Snead's assessment, a central problem with expressive individualism is that it offers an account of the human person that "cannot make sense of the fully lived reality of human *embodiment*, with all that entails".[8] Encounters with risk, dependency, and relationality are all aspects of human experience mediated through bodies, yet an anthropology premised on expressive individualism cannot render such experiences sensible or meaningful. In fact, if anything, such experiences only inhibit the free expressive autonomy of persons. They are points of vulnerability that puncture autonomy. Posthumanism likewise cannot provide an account of the ways in which embodied experiences such as these, which carve into the freedom of life, might at the same time contribute to life's moral density. In the end, the body remains a problem. Reducing the body to a site for imposing will and achieving authenticity has led to the evisceration of its moral significance. This is not only the case with posthuman thought. The evasion of the body might be seen as endemic to late modern culture, especially its biotechnological aspirations. Within a moral universe informed by the anthropology of expressive individualism, the human person is characterized not by gratuity and vulnerability but the drama of being and becoming into an open future. What then does it mean to be a dignified human? No coherent answer can be provided.

[7] C. SNEAD, *What It Means To Be Human: The Case for the Body in Public Bioethics*, Cambridge, 2020, 86.

[8] *Ivi*, 124.

Breve storia della genetica: cronologia, concetti, temi

Ilaria Ampollini*

A BRIEF HISTORY OF GENETICS: CHRONOLOGY, CONCEPTS, AND THEMES

ABSTRACT: The history of genetics, since its origin, has transcended the scientific dimension, interweaving social, cultural, and political contexts. This brief contribution aims both to offer a chronological overview of scientific achievements in the field of genetics, starting from the pivotal work of Mendel and Darwin, and to outline concepts and themes that have emerged over time. It will show how the history of genetics allows us to reflect on some peculiar dynamics of the history of scientific thought, such as the evolution of scientists' image, its relationship with society, the birth of new forms of cooperation (from the small lab to Big Science), and a constant, intense dialogue among the different social actors. All these elements still strongly characterize genetics today and investigating their historical roots will help us understand their nature and *raison d'être*.

KEYWORDS: Chronology; concepts; society; genetics; history

1. Introduzione

La storia degli studi, delle ipotesi e delle scoperte nel campo della genetica non inerisce solo alla sfera dello sviluppo della scienza *stricto sensu*. È così per qualsiasi cambiamento di paradigma[1] o nuova teoria, certo; tuttavia, nella nascita e nell'affermarsi delle teorie genetiche, gli intrecci tra l'evoluzione del pensiero scientifico e il contesto storico, sociologico e culturale sono particolarmente evidenti, ricorrenti e, in ultima istanza, meritevoli d'attenzione. È ovvio che questo legame tra produzione scientifica e contesti si nutre in questo caso soprattutto della prossimità del tema al significato della vita umana, della sua origine e del suo futuro. L'emergere di nuovi concetti, come quello di "carattere ereditario", o di "gene", sul finire dell'Ottocento, ha costretto a ripensare il passato dell'uomo, la sua evoluzione, il suo rapporto con le altre forme viventi e le sue specificità. Il dialogo, tra ciò che è avvenuto e avviene nei laboratori e nei luoghi di ricerca e ciò che se ne situa invece al di fuori, nell'ambito della genetica è ad oggi più intenso che mai, come dimostrano i dibattiti epistemologici, filosofici, etici, politici e giuridici sollecitati dalle nuove frontiere dell'editing genomico, delle biobanche e via dicendo.

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[1] T. KUHN, *The Structure of Scientific Revolutions*, Chicago, 1962.

ISSN 2284-4503

Il concetto stesso di gene, e con esso quello di genetica e quello di DNA, è entrato così prepotentemente a far parte della nostra cultura[2] e del nostro modo di pensare, che vari autori[3], partendo dall'ormai classico articolo di Star e Griesemer del 1989, ne hanno parlato come di un calzante esempio di "boundary object". Gli "oggetti di confine" sono «both plastic enough to adapt to local needs and the constraints of the several parties employing them, yet robust enough to maintain a common identity across sites. […] They have different meanings in different social worlds but their structure is common enough to more than one world to make them recognizable, a means of translation»[4]. È piuttosto immediato comprendere perché un "oggetto" come la struttura a doppia elica dell'acido desossiribonucleico e un termine come quello di "gene" si prestino facilmente a questa definizione. Questo, tuttavia, è già in parte un punto d'arrivo, un aspetto della questione che riconosciamo facilmente nella sensibilità e nella rielaborazione tipiche della nostra contemporaneità.

Vale allora la pena dare uno sguardo a come gene e genetica si sono costituiti, scientificamente e socialmente, nel tempo, partendo dagli studi di Mendel e fermandoci alla fine del Novecento, quando è stato lanciato lo *Human Genome Project*. In un secondo momento, andremo a guardare come si è modificato nel tempo il concetto di gene e come se ne sono costituiti altri, prima diffusi e poi superati, come quello di razza e di eugenetica. Metteremo infine in evidenza contesti e tematiche centrali nel dispiegarsi del pensiero scientifico, per i quali lo sviluppo della genetica offre numerosi spunti: si pensi ad esempio alla nascita dei laboratori in senso moderno e al cambiamento nelle modalità di collaborazione, oppure alla presenza femminile e ancora all'immagine dello scienziato nel suo rapporto con la società.

2. Da Charles Darwin al Progetto Genoma: una panoramica cronologica

Solitamente, le radici della storia della genetica e degli studi sull'ereditarietà dei caratteri si fanno risalire all'inglese Charles Robert Darwin (1809-1882) e al ceco Gregor Johann Mendel (1822-1884)[5]. Certo simili tematiche, di cui troviamo traccia già in autori classici come Aristotele o Democrito, hanno da sempre accompagnato la storia del pensiero filosofico e scientifico. A partire dalla prima età moderna, le riflessioni circa l'origine e l'evoluzione delle specie e i meccanismi di riproduzione si intensificarono notevolmente, producendo contributi fondamentali che di fatto prepararono la strada ai tanti passaggi cruciali che segnarono il XIXesimo secolo. Così le teorie di Darwin e Mendel, è bene

[2] Ad esempio, D. NELKIN, M. LINDEE, *The DNA Mystique: The Gene as a Cultural Icon*, Ann Arbor, 2004, II.

[3] M. BUCCHI, *Science and the Media. Alternative Routes in Scientific Communication*, London/New York, 1998, 30-32; H.J. RHEINBERGER, *Gene Concepts: Fragments from the Perspective of Molecular Biology*, in P.J. BEURTON, R. FALK, H.J. RHEINBERGER (eds.), *The Concept of the Gene in Development and Evolution: Historical and Epistemological Perspectives*. Cambridge, 2000, 219-239; E. PARTHENIA SHEA, *How the Gene Got Its Groove: Figurative Language, Science, and the Rhetoric of the Real*, 2008, cap. 5.

[4] S.L. STAR, J.R. GRIESEMER, *Institutional Ecology, Translations and Boundary Objects: Amateurs and Professionals in Berkeley's Museum of Vertebrate Zoology (1907-39)*, in *Social Studies of Science*, 19, 3, 1989, 393.

[5] La ricostruzione cronologica si è basata in particolare sui seguenti testi: P.S. AGUTTER, D.N. WHEATLEY, *Thinking about Life. The History and Philosophy of Biology and Other Sciences*, Dordrecht, 2008; P.J. BOWLER, J.V. PICKSTONE, *The Cambridge History of Science. Vol. 6 – Modern Life and Earth Sciences*, Cambridge, 2008; e sul più risalente P. ROSSI (a cura di), *Storia della Scienza e della Tecnica*, Torino, 1988, vol. II, tomo II; vol. III, tomi I e II. I capitoli sulla biologia sono firmati da B. FANTINI. Altre fonti sono di volta in volta specificate.

ricordarlo, furono precedute da lavori essenziali, come la teoria evoluzionistica elaborata da Jean-Baptiste Lamarck (1744-1829), o come gli studi sui caratteri ereditari condotti dagli ibridatori e da coltivatori e zootecnici tramite ricerche empiriche, che si moltiplicarono già nella seconda metà del Settecento[6]. Nello stesso tempo, è innegabile che furono proprio Darwin e Mendel a introdurre metodologie, contenuti e proposte interpretative che avranno un ruolo centrale per lo sviluppo delle conoscenze nei decenni successivi –anche se, ribadiamolo fin da ora, l'opera di Mendel sarà prima dimenticata e poi recuperata all'alba del Novecento, a differenza di quella darwiniana, che fin da subito si inserì con forza nei dibattiti dell'epoca.

Dell'importante produzione di Darwin, si ricordi qui l'imprescindibile *Origin of the Species*, pubblicato nel 1859: un'opera uscita dopo una lunga gestazione[7] e frutto dei viaggi che l'inglese compì a bordo della HMS Beagle, durante i quali ebbe l'opportunità di raccogliere una impressionante quantità di dati circa numerose specie animali e fossili. Procedendo per generalizzazione induttiva, Darwin arrivò a concepire una teoria dell'evoluzione basata sul concetto di variazioni casuali ed ereditabili, sulle quali l'azione della selezione naturale interviene a favore dell'organismo più adatto in un dato ambiente. Darwin avanzò anche una proposta circa il meccanismo di eredità delle variazioni, cui diede il nome di pangenesi, e che formulò nel testo *Variation of animals and plants under domestication* (1868). Secondo questa ipotesi, ogni parte del corpo produce dei corpuscoli, i *pangeni*, che confluiscono nelle cellule uovo e da qui partecipano del meccanismo riproduttivo.

Mendel basò invece le sue teorie su un'ampia serie di esperimenti che condusse tra il 1856 e il 1863, a Brünn, dove era entrato nel monastero agostiniano e dove insegnava come supplente nelle scuole secondarie. Basandosi sugli incroci di generazioni di *Pisum sativum*, la comune pianta di pisello, Mendel studiò la trasmissione e la ricombinazione dei caratteri ereditari nella discendenza. Alla luce degli incroci ottenuti, ipotizzò l'esistenza di due fattori, uno proveniente dal padre e uno dalla madre, laddove uno dei due poteva "nascondere" l'altro. Della metodologia messa in campo da Mendel, quello che più è rilevante non è solo l'utilizzo di un approccio quantitativo e sperimentale, ma anche, e soprattutto, l'applicazione del calcolo matematico e statistico ai risultati raccolti.

Ma come mai l'opera di Mendel passò fondamentalmente sotto silenzio? Sono state fatte diverse ipotesi a riguardo[8], ma nessuna sembra riuscire a spiegare in modo esaustivo la sostanziale indifferenza della comunità scientifica dell'epoca verso le conclusioni che Mendel presentò alla Società di Scienze Naturali di Brünn e che poi pubblicò negli Atti. Qualunque sia il motivo, bisognerà aspettare l'inizio del Novecento, quando Hugo De Vries (1848-1935) e Carl Erich Correns (1864-1933) recuperarono i risultati del monaco ceco, che solo da allora iniziarono ad avere fortuna e a incontrare conferme sperimentali e apprezzamenti generali. È a questo punto che entra in scena un volto cruciale per la storia della genetica: l'inglese William Bateson (1861-1926), che fu il principale sostenitore della teoria mendeliana e contribuì fortemente alla sua diffusione in terra anglofona.

[6] Una sintesi si trova anche in E. MAYR, *Storia del pensiero biologico* (ed. or. *The Growth of Biological Thought. Diversity, Evolution and Inheritance*, Cambridge 1982), Torino, 2011, III ed., vol. I, 589-599.

[7] J. VAN WYHE, *Mind the Gap: Did Darwin Avoid Publishing his Theory for Many Years?*, in *Notes and Records of the Royal Society of London*, 61, 2, 2007, 177-205.

[8] Si veda P. ROSSI (a cura di), *Storia della Scienza e della Tecnica*, vol. II, tomo II, 924-928.

Con i nuovi studi, divenne fondamentale chiedersi se i fattori mendeliani fossero delle realtà fisiche – e se sì, dove si situassero – e come avvenisse la trasmissione di un certo fattore dai genitori alla prole. Acquisì dunque rilevanza l'incontro tra la sperimentazione nel campo dell'agro-zootecnica, che aveva da sempre guardato alla trasmissione di caratteri, e le ricerche nel campo della citologia, che permettevano di comprendere sempre meglio la struttura e la composizione delle cellule.

In realtà, fin dalla seconda metà dell'Ottocento si sapeva dell'esistenza sia del DNA, se pur chiamato in modo diverso, sia dei cromosomi. Era stato lo svizzero Friedrich Miescher, nel 1869, a isolare il DNA, durante una serie di esperimenti sui leucociti: all'interno dei globuli bianchi, Miescher aveva individuato un composto ricco di fosforo e lo aveva chiamato nucleina, poiché si era accorto che si trovava nel nucleo cellulare. Quasi vent'anni più tardi, Albrecht Kossel (1853-1927), premio Nobel nel 1910, mostrò che la nucleina si componeva di una parte proteica e di una parte non proteica (l'acido nucleico). Procedendo con l'analisi degli acidi nucleici, identificò l'adenina, la citosina, la guanina, la timina e l'uracile. Al tempo, tuttavia, non era certamente possibile avanzare ipotesi circa un coinvolgimento del DNA nei meccanismi di trasmissione dei caratteri ereditari.

Un simile discorso vale per i cromosomi: scoperti nel 1878 da Walther Flemming (1843-1905), che studiò anche il processo di divisione cellulare, cui diede egli stesso il nome di mitosi, e chiamati così da Heinrich Wilhelm Gottfried von Waldeyer Hartz (1836-1921), furono messi in relazione con la trasmissione dei caratteri ereditari soltanto anni dopo. E, per inciso, bisognerà aspettare il 1955 per conoscere con esattezza il numero di cromosomi del cariotipo umano, grazie al lavoro di Joe-Hin Tjio (1919-2001) e Albert Levan (1905-1988), di formazione citologi vegetali.

Tra i primi a ipotizzare che i fattori mendeliani si localizzassero fisicamente su queste strutture filiformi, ci furono William A. Cannon (1870-1958) e Walter Sutton (1877-1916), per il quale fu fondamentale la collaborazione, presso la Stazione zoologica di Napoli, con Theodor Boveri (1862-1915), che aveva già raggiunto risultati importanti. Sutton, nel 1902, lavorando sui cromosomi della *Brachystola magna* (cavalletta), si accorse che il numero di cromosomi si dimezzava durante la meiosi e che ogni spermatozoo o cellula uovo riceveva dunque un solo cromosoma per ogni coppia. Nell'articolo *The Chromosomes in Heredity*, uscito nel 1903 sul *Biological Bulletin*[9], Sutton dimostrò la sostanziale coerenza dei risultati ottenuti con le conclusioni cui era arrivato Mendel e ipotizzò che i cromosomi fossero la base fisica delle leggi mendeliane dell'eredità.

Fu l'inizio di intense ricerche sui cromosomi e sulla localizzazione dei fattori mendeliani, che videro l'interesse da parte di molteplici studiosi e gruppi di ricerca: è proprio in questo passaggio cruciale, epistemologico prima ancora che sperimentale, che si identifica la fine della genetica formale e l'inizio di quella che può essere chiamata genetica dei cromosomi. Dei numerosi passi avanti realizzati durante i decenni successivi, occorre almeno ricordare i lavori di Nettie Y. Stevens (1861-1912), grazie ai quali venne identificato il cromosoma maschile Y: la scoperta fu pubblicata nella celebre opera[10] del 1905 *Studies in spermatogenesis with especial reference to the accessory chromosome* e

[9] W. SUTTON, *The Chromosomes in Heredity*, in *Biological Bulletin*, 4, 5, 1903, 231-251.

[10] N. M. STEVENS, *Studies in Spermatogenesis with Especial Reference to the "Accessory Chromosome"*, Washington D.C., 1905, Vol I. *Cfr*. S. G. BRUSH, *Nettie M. Stevens and the Discovery of Sex Determination by Chromosomes*, in *Isis*, 69, 1978, 162-72.

ISSN 2284-4503

faceva seguito ai risultati cui era giunto Erwin C. McClung (1870-1946), che aveva ricondotto la determinazione del sesso al solo cromosoma X.

Un forte impulso alle nuove linee di ricerca venne inoltre da Thomas H. Morgan (1866-1945), Premio Nobel per la Medicina nel 1933, e dal suo gruppo di ricerca, le cui sperimentazioni si concentrarono sull'organismo della *Drosophila melanogaster* (moscerino della frutta). Inizialmente Morgan, oltre a essere propenso per una teoria di tipo epigenetico, era anche scettico rispetto agli studi di Sutton ed era più incline a credere che fosse il citoplasma, e non tanto il nucleo (e dunque i cromosomi), a svolgere un ruolo di rilievo nell'eredità. Dopo aver studiato la mutazione "occhi bianchi" sulla Drosophila e averne analizzato la trasmissione alla progenie, Morgan rivide completamente le sue posizioni. Oltre a sostenere che i geni si trovassero fisicamente sui cromosomi, spiegò anche il fatto che alcuni "fattori mendeliani" si ricombinassero durante la meiosi con le teorie di un citologo belga, Frans Alfons Janssens (1865-1924), che per primo nel 1904 aveva descritto il fenomeno del crossing-over. Janssens aveva infatti rilevato come, nel corso della meiosi, i membri delle coppie di cromosomi si arrotolano l'uno con l'altro, favorendo lo scambio di segmenti di filamento. Quanto più due fattori solitamente associati (ad esempio ali e colore occhi) finiscono per dividersi, tanto più la loro posizione sui cromosomi deve essere lontana. Proprio questa considerazione fu alla base dei primi tentativi di mappatura intrapresa negli anni successivi dal gruppo di Morgan, in particolare da Alfred H. Sturtevant (1891-1970).

L'ormai assodata variabilità dei geni e la presenza di fenomeni di ricombinazione intensificò l'interesse per le mutazioni, interesse che portò con sé una stimolante questione metodologica: come fare per studiare in modo sistematico un fenomeno che è di per sé casuale e imprevedibile? Nacquero così i tentativi di indurre le mutazioni in laboratorio. Uno degli scienziati che più lavorò al problema proveniva dal gruppo di Morgan: si tratta di Herman Joseph Muller (1890-1967), Premio Nobel in Medicina nel 1946, che dimostrò come l'utilizzo di massicce dosi di raggi X potesse indurre delle mutazioni nelle cellule uovo e spermatiche[11]. Muller sostenne inoltre, a differenza di molti suoi colleghi, che le mutazioni indotte con questo metodo fossero le stesse che avvengono per via naturale[12]. Solo più avanti nel tempo iniziarono ad essere utilizzati mutageni di tipo chimico.

Gli studi sulle mutazioni rappresentarono un'altra, fondamentale linea di ricerca, che si intrecciò con le ipotesi circa il ruolo svolto dai geni nel controllo e nella regolazione delle reazioni metaboliche. Uno dei primi esempi in questo senso precede i risultati di Muller: fu tra il 1902 e il 1908, infatti che Archibad Garrod (1857-1936) osservò che alcune malattie metaboliche, come dell'alcaptonuria, l'albinismo, la cistinuria e la pentosuria, vengono trasmesse secondo le leggi di Mendel e ipotizzò dunque che la loro causa fosse da ricercarsi in una mutazione di un fattore ereditario. Sono in effetti tutte malattie determinate dalla mancanza di uno specifico enzima. Le ipotesi di Garrod torneranno di forte attualità negli anni '40, quando George Beadle (1903-1989) e Edward Tatum (1909-1975) approdarono alla celebre formulazione un gene-un enzima (divenuta poi un gene-una catena polipeptidica, laddove, come sappiamo, una tripletta di basi codifica per un amminoacido) ed inferirono dunque che le due funzioni principali dei geni fossero l'auto-replicazione e la produzione di enzimi.

[11] H.J. MULLER, *Artificial Transmutation of the Gene*, in *Science*, 66, 1927, 84-87; ID., *The Problem of Genic Modification*, in *Proceedings of the 5th International Congress* 1, 1928, 234-260.

[12] ID., *Artificial Transmutation of the Gene*, 84-87.

Lo sviluppo[13] negli anni '30 delle analisi dei cromosomi umani, fino ad allora poco studiati, reso possibile dall'evoluzione delle tecniche citogenetiche, aprì la strada alla genetica umana e con essa all'intensificarsi delle scoperte delle malattie su base genetica. Negli anni '40 fu la volta della talassemia, la cui origine genetica fu dimostrata da due ricercatori italiani, Ida Bianco (1917-2006) e il marito Ezio Silvestroni (1905-1990), e da James V. Neel (1915-2000), che proseguì i propri studi interessandosi anche dell'anemia falciforme. Com'è noto, fu poi Linus Pauling (1901-1994), successivamente premio Nobel per la Chimica e per la Pace, ad approfondire nel 1949 le proprietà fisiche dell'emoglobina in caso di anemia ed infine a confermare il legame tra geni e sintesi proteiche. Nel decennio successivo l'attenzione si allargò alle malattie determinate da anomalie cromosomiche. Nel 1959, per esempio, Jérôme Lejeune (1926-1994), contando sulla preziosa collaborazione di Marthe Gauthier (1925), specializzata nell'analisi delle colture cellulari (e usando la tecnica citologica di Tjio e Levan), riuscì ad osservare che la sindrome di Down era collegata non all'assenza di un cromosoma, ma alla presenza di un cromosoma in più.

Nel frattempo, il cosiddetto Gruppo del Fago, una rete informale di collaborazione costituita, tra gli altri, dal fisico di formazione Max Delbrück (1906-1981), Alfred D. Harshey (1908-1997) e Salvador Luria (1912–1991), si occupò di un nuovo problema divenuto centrale, ossia cercare di capire se il materiale genetico fosse trasportato dalle proteine o dal DNA, entrambi presenti nel nucleo. Gli esperimenti, realizzati utilizzando un batteriofago, di cui erano state caricate radioattivamente le parti di fosforo, arrivarono a raccogliere evidenze del fatto che fosse il DNA a contenere l'informazione ereditaria. Si verificò infatti che il virus rilasciava all'interno del batterio, allo scopo di "colonizzarlo", non le proteine, ma solo il DNA. La scoperta fruttò a Delbrück, Harshey e Luria il Premio Nobel per la Medicina nel 1969[14].

Le crescenti ricerche circa le proprietà dell'Acido desossiribonucleico consentirono, nel 1953, di scoprirne la struttura a doppia elica: com'è noto, il lavoro, realizzato da James Watson (1928) e Francis Crick (1916-2004), non sarebbe mai stato possibile senza gli studi di Maurice Wilkins (1916-2004) e Rosalind Franklin (1920-1958). A questo punto, era necessario spiegare non solo i meccanismi di auto-replicazione del DNA, ma anche in che modo avvenisse il controllo della sintesi proteica: l'identificazione dell'RNA messaggero non tardò ad arrivare. In un articolo[15] del 1961, pubblicato su *Nature*, infine, Crick, assieme a Leslie Barnett (1920-2002), Sydney Brenner (1927-2019) e Richard Watts-Tobin (1934-), espose la decifrazione del codice genetico: la successione di basi andava letta a triplette, poiché ogni tripletta (o codone) codifica un amminoacido.

Negli anni successivi, i passi avanti furono innumerevoli e determinanti. Sarebbe troppo complesso ripercorrere tutte le tappe: si ricordi, a titolo esemplificativo e molto brevemente, la scoperta, nel

[13] Non c'è qui lo spazio per parlare della sintesi evoluzionistica, che vide confluire l'evoluzionismo darwiniano e la teoria mendeliana in uno studio delle popolazioni che teneva conto dei più recenti risultati nel campo della genetica e degli strumenti della statistica. Ne sono considerati gli iniziatori John Haldane (1892-1964), Sewall Wright (1889-1988) e Ronald Fisher (1890-1962). Per una ricostruzione storica firmata da uno dei protagonisti del dibattito, si rimanda a E. MAYR, *op. cit.*, vol. II.

[14] Le motivazioni si trovano sul sito www.nobelprize.org/prizes/medicine/1969/summary/, consultato il 20 Gennaio 2021.

[15] F. CRICK, L. BARNETT, S. BRENNER, R. WATTS-TOBIN, *General Nature of the Genetic Code for Proteins*, in *Nature*, 192, 1961, 1227-1232.

1977, dell'esistenza di introni ed esoni, e dunque della presenza di DNA non codificante; i primi passi nelle biotecnologie; e ancora la localizzazione, nel 1983, del primo marker polimorfico legato a una malattia genetica, ovvero quello relativo alla Coréa di Huntington, individuato sul cromosoma 4[16].
L'accumularsi di conoscenze sempre più precise e complesse confluì, sul finire degli anni Ottanta, nella nascita dello *Human Genome Project*. A fronte dei considerevoli costi richiesti dal progetto, il primo di così grande portata nel campo delle scienze della vita, non mancarono critiche e interrogativi. Di fatto, il sequenziamento completo del genoma umano ha permesso di aprire definitivamente una nuova fase nella storia della genetica, di cui oggi vediamo solo i primi effetti, incentrata sulla possibilità di intervenire direttamente sui geni, per esempio per prevenire o curare malattie tramite la modifica del DNA.

3. Concetti in evoluzione

Come si intravede da questa sintetica e certo non esaustiva ricostruzione cronologica, la storia della genetica è segnata dall'emergere e dall'avvicendarsi di nuovi concetti, i cui significati cambiano di continuo, intrecciandosi di volta in volta con questioni filosofiche, storiche, sociologiche e culturali. Ad essere centrale è ovviamente il concetto di gene; non è tuttavia l'unico a meritare attenzione. Altri concetti, come quello di ereditarietà, di "razza", di eugenetica ed epigenetica, hanno rivestito e rivestono tutt'oggi un ruolo fortemente di rilievo, dimostrando, se mai ce ne fosse bisogno, una volta in più come e quanto le ricerche sul DNA, l'evoluzione delle specie e i meccanismi di riproduzione cellulari abbiano catalizzato, in passato come ora, riflessioni e dibatti che hanno coinvolto in eguale misura mondo della ricerca e società.
Come evidenziato da svariati lavori[17], il concetto di gene ha assunto, nella storia della genetica, significati profondamente differenti, non solo relativamente alle fasi temporali che hanno caratterizzato gli studi sull'ereditarietà, ma anche a seconda dell'approccio epistemologico dei ricercatori che di volta in volta se ne sono occupati.
Il termine gene fu introdotto[18] nel 1908 da Wilhelm Johannsen (1857-1927), botanico danese, e andò presto a sostituire una serie di vocaboli che erano stati utilizzati fino ad allora, dal *Merkmal*, ossia "carattere", "fattore", di Mendel, all'*Anlage* −unità− dell'olandese De Vries e ancora lo *Unit-character* proposto da Bateson e traducibile come "carattere unitario". Il problema risiedeva nella confusione semantica che questi termini non riuscivano a risolvere: potevano riferirsi al carattere visibile oppure alla corrispondente base ereditaria. Proprio per questo motivo, sempre Johannsen introdusse la chiara distinzione tra "fenotipo" e "genotipo". La proposta di ricorrere al termine "gene" era un richiamo

[16] J.F. Gusella et al., *A polymorphic DNA marker genetically linked to Huntington's disease*, in *Nature*, 306, 1983, 234-238.
[17] Per una trattazione approfondita: E.F. Keller, *The Century of the Gene*, Cambridge, 2000; P.J. Beurton, R. Falk, H.J. Rheinberger, *The Concept of the Gene in Development and Evolution: Historical and Epistemological Perspectives*. Cambridge, 2000; P. Portin, *The Concept of the Gene: Short History and Present Status*, in *The Quarterly Review of Biology*, 68, 2, 1993, 173-223; E.A. Carlson, *Defining the Gene: An Evolving Concept*, in *American Journal of Human Genetics*, 49, 1991, 475-487.
[18] Curiosamente, già quattro anni prima, in una lettera privata, William Bateson aveva utilizzato il termine "genetica", che tuttavia non si diffuse se non dopo l'introduzione da parte di Johannsen di "gene".

al "pangene" con cui Darwin e De Vries avevano indicato le particelle ereditarie. Questo cruciale cambiamento lessicale portò con sé questioni non solo di tipo semantico, ma anche epistemologico, scientifico e filosofico: il primo problema che infatti ora emergeva con rinnovata evidenza era quale fosse il rapporto tra gene e carattere, quali meccanismi chimici e biologici li mettessero in relazione, ma soprattutto quale fosse la natura del gene. Se il carattere fenotipico era ciò che "si vedeva" empiricamente, come andava inteso il gene? Gli si poteva attribuire una realtà materiale o piuttosto andava considerato come un'unità di calcolo? L'approccio formale, termine con cui si è soliti indicare la fase pre-molecolare, o classica[19], della storia della genetica, di fatto non si preoccupava di cosa fosse, concretamente e chimicamente, un gene. Lo stesso Johannsen non era interessato a rispondere alla domanda, come questo passaggio del suo fondamentale testo *Elemente der exakten Erblichkeitslehre*[20] dimostra: «La parola gene è completamente scevra da qualsiasi ipotesi; esprime solo il fatto stabilito, che almeno molte proprietà di un organismo sono condizionate da "condizioni", "fondamenti", "disposizioni" particolari, separabili e quindi indipendenti»[21]. Nella terza edizione del 1926, Johannsen avrà a sottolineare la dimensione concreta del gene[22] – «[i] geni sono realtà, non concezioni ipotetiche» –, salvo poi concludere ribadendo che i geni sono «unità di calcolo, espressioni di realtà di natura sconosciuta»[23]. Nonostante le possibili, apparenti contraddizioni, è piuttosto chiaro che per Johannsen è importante affermare che i geni esistono, che "ci sono"; meno importante è stabilire "cosa sono", perché non funzionale alla spiegazione dei meccanismi di ereditarietà.

Già la fase citogenetica implica un cambiamento, dal momento che con Sutton e poi gli studi di Morgan e del suo gruppo di ricerca l'interesse si sposta sui cromosomi[24], sulla loro costituzione e sulla possibile localizzazione dei geni. Relativamente a questo periodo, resta fondamentale il testo di Hermann Muller, letto durante una conferenza nel 1926, in cui il genetista statunitense ribadì la propria convinzione che il gene fosse *la* base della vita e non soltanto *una* base della vita. Per inciso, Muller sottolineò più volte il ruolo chiave delle mutazioni e, più di tutto, dell'ereditarietà delle mutazioni, laddove era essenziale considerare mutazioni e ereditarietà come componenti chiave dello stesso meccanismo, e non come due processi separati[25].

Un ulteriore superamento si verifica con l'ingresso nella fase molecolare – che Portin chiama neoclassica –, e dunque con l'individuazione della struttura a doppia elica del DNA e dei meccanismi di codifica delle catene polipeptidiche. Gli anni che seguono il 1953 mettono duramente alla prova il concetto di gene come unità di informazione: basti pensare alla scoperta degli introni fatta nel 1976, che dimostrò la presenza di materiale non codificante, e dunque apparentemente inutilizzato e inutilizzabile, all'interno di ogni gene, ma anche a tutte le successive sperimentazioni che hanno portato

[19] P. PORTIN, *op. cit.*, 175-179.
[20] W. JOHANNSEN, *Elemente der exakten Erblichkeitslehre*, Jena, 1909.
[21] In N. ROLL-HANSEN, *Commentary: Wilhelm Johannsen and the problem of heredity at the turn of the 19th century*, in *International Journal of Epidemiology*, 43(4), 2014, 1007-1013. Nello specifico, 1011.
[22] W. JOHANNSEN, *Elemente der exakten Erblichkeitslehre,* Jena, 1926, III ed.
[23] N. ROLL-HANSEN, *op. cit.*, 1011.
[24] E.A. CARLSON, *The Drosophila Group: The Transition from the Mendelian Unit to the Individual Gene*, in *Journal of the History of Biology*, 7, 1, 1974, 31-48.
[25] E. A. CARLSON, *The Drosophila Group: The Transition from the Mendelian Unit to the Individual Gene*, *cit*., 31. Il testo di Muller a cui si fa riferimento è H. J. MULLER, *Variation Due to Change in the Individual Gene*, in *The American Naturalist*, 56, 642, 1922, 32-50.

ad intervenire direttamente sul gene per modificarlo. Quanto più il gene è stato indagato, tanto più il suo concetto è andato incontro ad una complessità difficile da maneggiare. Sono molto efficaci in questo senso le parole di Portin, che già trent'anni fa scriveva:

«[Due to] the discoveries of repeated genes, split genes, nested genes, overlapping genes, transposable genes, alter-native splicing, multiple and complex promoters, enhancers and silencers, downstream signals, internal control signals, proteolytic cleavage of translation products and other types of protein processing, editing of primary transcripts, the special case of immunoglobin genes, and the detailed analysis of gene complexes […], our comprehension of the nature of the gene entered a dramatic new phase. Paradoxically, in spite of the new, sometimes over-whelming, concreteness of our comprehension of the gene as a result of DNA technology, we seem to be left with a rather abstract and generalized concept of the gene that has quite different significances in different contexts»[26].

Proprio quest'ultimo passaggio, in cui Portin sostiene che il concetto di gene "ha significati piuttosto diversi in differenti contesti", ci riporta all'idea contemporanea del gene come "oggetto di confine", che attraversa ambiti e contesti, nei quali di volta in volta assume valenze semantiche rinnovate. In particolare, non si può non sottolineare come oggi il "gene" non sia in alcun modo un concetto ad appannaggio esclusivo del linguaggio scientifico, ma appartenga plasticamente alla realtà sociale, culturale, nonché giuridica, politica ed economica.
D'altronde, è tipico della storia della genetica, come abbiamo già avuto modo di ribadire, incrociare dimensioni eterogenee che esulano da quella meramente scientifica. Un altro concetto ce ne dà una chiara dimostrazione, ed è il concetto di eugenetica[27].
Ben prima delle tristi e note derive a cui condussero il regime nazista e fascista, il termine eugenetica era stato coniato nel 1883 da Francis Galton (1822-1911) [28], che lo avrebbe poi definito come «the study of the agencies under social control that may improve or impair the racial qualities of future generations either physically or mentally»[29]. Agli inizi del Novecento, i programmi di eugenetica si moltiplicarono, come testimonia il susseguirsi di convegni, società e riviste: ne sono un esempio l'americano *Eugenics Record Office*, l'inglese *Eugenics Society*, dedicata alla divulgazione, e la *Eugenics Review*[30]. L'idea di base era quella di arrivare a "migliorare" la specie umana, eliminando fenomeni come l'alcolismo, la criminalità, la "degenerazione morale" e addirittura l'erotismo innato: il programma poggiava su un assunto di fondo e cioè che tutti questi comportamenti fossero caratteri fenotipici determinati solo e soltanto da un unico gene (l'ambiente o situazioni di degrado non venivano presi in considerazione) e che dunque sarebbe stato facili eliminarli, per esempio procedendo con campagne di sterilizzazione mirata. Scienziati del calibro di Muller o Morgan vi aderirono e negli Stati Uniti degli anni '30 si arrivò a mettere in atto una serie di sterilizzazioni forzate. Con la salita al potere di Hitler e l'utilizzo che il Regime Nazista fece di queste teorie, unitamente alla graduale con-

[26] P. PORTIN, *op. cit.*, 174.
[27] Si faccia riferimento a A. BASHFORD, P. LEVINE (eds.), *The Oxford Handbook of the History of Eugenics*, Oxford, 2010. Si consideri anche F. CASSATA, *Molti, sani e forti. L'eugenetica in Italia*, Torino, 2006.
[28] F. GALTON, *Inquiries into Human Faculty and its Development,* London, 1883.
[29] ID., *Memories of my Life*, London, 1908, 321.
[30] L. BLAND, L.A HALL, *Eugenics in Britain: The View from the Metropole*, in A. BASHFORD, P. LEVINE (eds.), *op. cit.*, 213-227.

sapevolezza che si trattava di un concetto privo di fondamento scientifico (presto si scoprì che ogni carattere fenotipico è in realtà determinato da più geni), negli ambienti scientifici l'opposizione al concetto di eugenetica divenne molto forte e si susseguirono critiche sferzanti. Il termine non fu più utilizzato, se non in senso negativo, e i primi programmi di genetica umana promossero più di tutto la difesa del patrimonio genetico umano e della sua variabilità, intesa come garanzia di sopravvivenza della specie[31].

Un percorso e un destino simile fu quello che incontrò il concetto di razza: le scoperte sull'estrema variabilità del DNA umano e sull'evoluzione della specie umana ne dimostrarono l'inattendibilità scientifica e furono la conferma definitiva dell'impossibilità di attuare una classificazione discreta e oggettiva su base genetica[32]. La critica al concetto proveniente dal mondo della ricerca di fatto si unì a una più ampia rivisitazione del termine, che gli avvenimenti politici e i cambiamenti sociali e culturali della seconda metà del Novecento avevano reso necessaria e irrimandabile.

Per alcuni termini che furono abbandonati con decisione, se ne citi almeno uno che fu invece introdotto nel 1942 e che ad oggi continua ad essere utilizzato. Si tratta del concetto di epigenetica, una branca recente della genetica, che si occupa di studiare come l'ambiente influenzi l'espressione genica, senza tuttavia alterare le sequenze del DNA.

4. Conclusioni: contesti, volti, dinamiche

La storia della genetica è un'incredibile lente di ingrandimento che ci consente di guardare da vicino come lavora e come ha lavorato la scienza nel corso del secolo scorso. Se ci soffermiamo infatti sui luoghi e sui protagonisti degli studi sul gene ne ricaviamo non solo un avvicendarsi di ipotesi, scoperte e metodologie, ma anche una ricca panoramica di contesti, luoghi e dinamiche socio-culturali.

Iniziamo col guardare ai due scienziati che, come detto, sono generalmente indicati come coloro che hanno dato l'impulso determinante alle ricerche sull'evoluzione e sull'ereditarietà dei caratteri sviluppatesi nel corso del Novecento: Mendel e Darwin. La loro biografia e il loro percorso intellettuale, sotto molti punti di vista differenti, ci permettono di aprire una finestra su cosa significasse essere un "uomo di scienza" nella seconda metà dell'Ottocento. Mendel è figlio di contadini e per proseguire gli studi entra in monastero, dove la nomina ad abate lo allontanerà di fatto dal proseguire i propri lavori scientifici; Darwin proviene invece da una ricca famiglia: figlio di un medico e nipote di un filosofo naturalista, può sostenere i propri viaggi e le proprie ricerche senza preoccupazioni economiche. Per entrambi, quella di scienziato non è una professione: è un'occupazione sviluppata seguendo le proprie inclinazioni e i propri interessi, per la quale non percepiscono alcun guadagno. Questo punto è cruciale nella storia del pensiero scientifico e nell'evolversi della figura dello scienziato, soprattutto nel suo rapporto con la società[33]. È infatti solo nel corso del Novecento, a fronte di una progressiva

[31] B. Fantini, in P. Rossi (a cura di), *Storia della Scienza e della Tecnica*, 1988, Vol. III, Tomo II, 793.

[32] L.L. Cavalli-Sforza, P. Menozzi, A. Piazza, *The History and Geography of Human Genes*, Princeton, 1994, 16-20.

[33] Sulle immagini dello scienziato nel corso della storia della scienza, si vedano i fondamentali lavori di Shapin, in particolare S. Shapin, *The Way We Trust Now: The Authority of Science and the Character of the Scientist*, in P. Hoodbhoy, D. Glaser, S. Shapin (eds), *Trust Me, I'm a Scientist*, London, 2004, 42-63; S. Shapin, "The Man of Science," in L. Daston, K. Park (eds), *The Cambridge History of Science. Vol. 3: Early Modern Science*, Cambridge,

istituzionalizzazione della scienza e degli ambiti in cui viene prodotta conoscenza, che emerge la figura dello scienziato di professione, che non fa più ricerca (solo) per mera vocazione, ma (anche) per lavoro, percependone un guadagno. Si tratta di un cambiamento paradigmatico, che ha prodotto non solo nuovi panorami del sapere scientifico, ma anche nuovi interrogativi e rielaborazioni da parte dei vari attori sociali. Non è dunque un caso che la scelta di Craig Venter (1946) di fondare la *Celera Genomics*, come risposta privata allo *Human Genome Project* abbia catalizzato dibattiti e riflessioni crescenti sulla liceità e la moralità della sua condotta[34]. Il volto di questo scienziato-imprenditore personifica il punto più alto di questa tensione continua tra la percezione, di origine risalente, dello scienziato come qualcuno dotato di una particolare levatura morale, che svolge i suoi studi nel pieno disinteresse economico, e l'emergere invece dello scienziato professionista che non solo persegue il guadagno come "effetto collaterale", ma anzi utilizza la ricerca scientifica come una vera e propria attività imprenditoriale.

E d'altronde, il fatto che l'oggetto di ricerca sia il genoma, e dunque l'essenza stessa della vita umana, non poteva non portare con sé intensi interrogativi etici, giuridici e filosofici, ben riassunti dalla domanda: *di chi è* il genoma[35]?

Un'altra dinamica cui la storia della genetica permette di accennare inerisce alla nascita del laboratorio in senso moderno. Se guardiamo infatti a Mendel e a Darwin, per quanto inseriti in una rete di contatti fondamentali per la loro formazione e per la ricezione dei loro lavori, è chiaro che siamo davanti a due uomini di scienza che si dedicano alle proprie ricerche perlopiù in solitaria. Dobbiamo andare avanti nel tempo per trovare collaborazioni più estese, continuative e finanche istituzionalizzate e per arrivare ai primi, veri e propri gruppi di ricerca nel campo delle scienze della vita. Uno dei primi ed anche dei più noti e più importanti laboratori nella storia della genetica è senza dubbio quello nato attorno alla figura di Morgan, conosciuto anche come "Gruppo della Drosophila", con riferimento al modello animale utilizzato, o "Gruppo delle mosche", dal nome con cui veniva chiamata la stanza in cui i ricercatori si riunivano. Qui, all'interno del Dipartimento di Zoologia della *Columbia University*, lavoravano insieme biologi come Muller, Sturtevant e Calvin B. Bridges (1889-1938)[36]. Un altro esempio è rappresentato dal celebre Gruppo del Fago: un gruppo informale in realtà, più che un vero e proprio laboratorio, che ruotava attorno alla figura di Delbrück, in cui giocò un ruolo chiave l'incontro tra scienziati di formazione diversa. Da questo gruppo nacque il *Phage Course*, una *summer school* presso il *Cold Spring Harbor Laboratory*, che fu fondamentale nella formazione di numerosi ricercatori.

Ancora più interessante è notare che, dalla dimensione tutto sommato circoscritta e ristretta del laboratorio, il raggio d'azione si è ampliato sempre di più, prima con l'intensificarsi delle collaborazioni

2006, 179-191; S. SHAPIN, *Figures de scientifiques*, in *Histoire des sciences et des savoirs, Vol. 3: Le siècle des technosciences (depuis 1914)*, Paris, 2015, 27-45.

[34] Per esempio, S. SHAPIN, *Figures de scientifiques*, 27-45; J. WITKOWSKI, *A life worth writing about*, in *Nature*, 449, 2007, 785-786.

[35] M. ANGRIST, R. M. COOK-DEEGAN, *Who Owns the Genome?*, in *The New Atlantis*, 11, 2006, 87-96.

[36] Si veda E. A. CARLSON, *The Drosophila Group: The Transition from the Mendelian Unit to the Individual Gene*, *op. cit.*. Sulla rilevanza delle specifiche dinamiche di laboratorio nella costruzione della conoscenza scientifica, non si può non considerare B. LATOUR, S. WOOLGAR, *Laboratory Life*: *The Construction of Scientific Facts*, Beverlt Hills, 1979; B. LATOUR, *Science in Action: How to Follow Scientists and Engineers Through Society*, Cambridge, 1987.

tra vari istituti di ricerca, poi con l'avvio dello *Human Genome Project*[37], una vera e propria finestra sulla *Big Science* che caratterizza il nostro secolo e che trova il suo corrispettivo, in altri ambiti, nell' Large Hadron Collider (LHC) del CERN di Ginevra o nella Stazione Spaziale Internazionale. L'*International Human Genome Sequencing Consortium* è arrivato a coinvolgere non solo Stati Uniti e UK, ma anche Germania, Giappone, Cina e Francia[38].

Né va dimenticato, infine, che anche la storia della genetica, come tutta la storia del sapere scientifico, è affollata di figure femminili che la memoria storica ci ha consegnato a fatica, nonostante i contributi fondamentali che hanno dato. Il nome più noto è certamente quello di Rosalind Franklin, ma non è la sola. Si pensi alla già citata Nettie Maria Stevens, a cui si deve la scoperta del cromosoma Y, oppure a Barbara McClintock, Premio Nobel per la Medicina e la Fisiologia nel 1983, che negli anni '50 scoprì i trasposoni (elementi genetici in grado di spostarsi) o ancora a Martha Chase, cui si deve l'esperimento cruciale con i batteriofagi che portò all'identificazione del DNA come portatore del materiale genetico: l'esperimento prende oggi il nome di esperimento *Harshey-Chase*, eppure di rado è ricordata la collaborazione della *Chase* con il Gruppo del Fago.

Tutte queste dinamiche, che la storia della genetica mette ben in luce, sono una finestra sul più ampio panorama dello sviluppo del pensiero scientifico. Certo la genetica è luogo d'osservazione privilegiato: molto ci dice del nostro passato e del nostro presente e molto continuerà a raccontare a chi un domani si guarderà indietro.

[37] La bibliografia è davvero ricca. Si veda a titolo d'esempio H. ZWART, *Human Genome Project: History and Assessment*, in *International Encyclopedia of Social & Behavioral Sciences*, Oxford, 2015, II ed., 311-317.

[38] Vedi www.genome.gov/human-genome-project, (consultato il 18 Gennaio 2021).

ISSN 2284-4503

Special issue

A brief history of genetics: Chronology, concepts, and themes

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ABSTRACT: The history of genetics, since its origin, has transcended the scientific dimension, interweaving social, cultural, and political contexts. This brief contribution aims both to offer a chronological overview of scientific achievements in the field of genetics, starting from the pivotal work of Mendel and Darwin, and to outline concepts and themes that have emerged over time. It will show how the history of genetics allows us to reflect on some peculiar dynamics of the history of scientific thought, such as the evolution of scientists' image, its relationship with society, the birth of new forms of cooperation (from the small lab to Big Science), and a constant, intense dialogue among the different social actors. All these elements still strongly characterize genetics today and investigating their historical roots will help us understand their nature and *raison d'être*.

KEYWORDS: Chronology; concepts; society; genetics; history

1. Introduction

The history of studies, hypotheses, and discoveries in the field of genetics is not only a matter of the development of science. This is true of any paradigm shift[1] or new theory, of course, but in the birth and emergence of genetic theories, the links between the evolution of scientific thought and the historical, sociological, and cultural context are particularly evident, recurrent, and ultimately worthy of attention. Obviously in this case, these links between scientific production and its contexts are nourished primarily by the proximity of the theme to the meaning of human life, its origin, and its future. The emergence of new concepts, such as "hereditary character" or "gene", at the end of the nineteenth century forced us to rethink humankind's past, its evolution, its relationship with other living forms, and its specificities. In the field of genetics, the dialogue between what has taken place and is taking place in laboratories and research centres and what lies outside them is now more intense than ever, as demonstrated by the epistemological, philosophical, ethical, political, and legal debates prompted by the new frontiers of genomic editing, biobanks, and so on.

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[1] T. KUHN, *The Structure of Scientific Revolutions*, Chicago, 1962.

ISSN 2284-4503

The very concept of the gene, as well as those of genetics and DNA, has become such an integral part of our culture[2] and way of thinking that various authors,[3] building on the classic article by Star and Griesemer in 1989, have referred to it as a fitting example of a "boundary object". Boundary objects are "both plastic enough to adapt to local needs and the constraints of the several parties employing them, yet robust enough to maintain a common identity across sites. [...] They have different meanings in different social worlds, but their structure is common enough to more than one world to make them recognizable, a means of translation".[4] It is quite easy to understand why an 'object' such as the double-helix structure of deoxyribonucleic acid and a term such as *gene* easily lend themselves to this definition. This, however, is already in some way a point of arrival, something that we easily recognise in the sensitivity and reinterpretation of our contemporary world.

It is therefore interesting to look at how genes and genetics have been scientifically and socially constituted over time, starting with Mendel's research and going to the end of the twentieth century, when the Human Genome Project was launched. In the second step, we will consider how the concept of the gene has changed over time and how other concepts, such as race and eugenics, were first widespread and then became outdated. Finally, we will highlight the contexts and themes central to the development of scientific thought, for which the evolution of genetics offers numerous opportunities: for example, the birth of laboratories in the modern sense and the changes in their collaboration, or women's contributions, or the image of the scientist in his or her relationship with society.

2. From Charles Darwin to the Genome Project: A chronological overview

The roots of genetics and the study of the inheritance of traits can usually be traced back to the Englishman Charles Robert Darwin (1809–1882) and the Czech Gregor Johann Mendel (1822–1884).[5] Certainly, such themes, which can already be found in classical authors such as Aristotle or Democritus, have always accompanied the history of philosophical and scientific thought. From the early modern age onwards, reflections on the origin and evolution of species and the mechanisms of reproduction intensified considerably, producing fundamental contributions that paved the way for the many crucial steps that marked the nineteenth century. Thus, Darwin's and Mendel's theories were preceded by essential works, such as the evolutionary theory developed by Jean-Baptiste Lamarck

[2] See, for instance, D. NELKIN, M. LINDEE, *The DNA Mystique: The Gene as a Cultural Icon*, Ann Arbor, 2004.

[3] M. BUCCHI, *Science and the Media: Alternative Routes in Scientific Communication*, London/New York, 1998, pp. 30-32; H.-J. RHEINBERGER, *Gene Concepts: Fragments from the Perspective of Molecular Biology*, in P. J. BEURTON, R. FALK, H.-J. RHEINBERGER (eds.), *The Concept of the Gene in Development and Evolution: Historical and Epistemological Perspectives,* Cambridge, 2000, pp. 219-239; E. PARTHENIA SHEA, *How the Gene Got Its Groove: Figurative Language, Science, and the Rhetoric of the Real*, 2008, chap. 5.

[4] S. L. STAR, J. R. GRIESEMER, *Institutional Ecology, 'Translations' and Boundary Objects: Amateurs and Professionals in Berkeley's Museum of Vertebrate Zoology (1907-39)*, in *Social Studies of Science*, 19 (3), 1989, p. 393.

[5] The chronological reconstruction was based in particular on the following texts: P. S. AGUTTER, D. N. WHEATLEY, *Thinking about Life: The History and Philosophy of Biology and Other Sciences*, Dordrecht, 2008; P. J. BOWLER, J. V. PICKSTONE, *The Cambridge History of Science, Vol. 6: Modern Life and Earth Sciences*, Cambridge, 2008; and the older P. ROSSI (ed.), *Storia della Scienza e della Tecnica*, Torino, 1988, vol. II, tome II; vol. III, tomes I & II. The chapters on biology are authored by B. FANTINI. Other sources are specified in the following.

(1744–1829) or the studies on hereditary traits carried out by hybridisers, cultivators, and zootechnicians through empirical research, which had already multiplied in the second half of the eighteenth century.[6] At the same time, it is undeniable that it was Darwin and Mendel who introduced the methodologies, contents, and interpretative proposals that were to play a central role in the development of knowledge in the decades that followed—although we should remember at this point that initially, Mendel's work was forgotten; it was then rediscovered at the dawn of the twentieth century, unlike Darwin's, which was immediately included in the debates of the time.

Of Darwin's important works, we recall here the indispensable *Origin of the Species*, published in 1859: a work that came out after a long gestation[7] and that was the result of the journeys that the Englishman made on board the HMS *Beagle*, during which he had the opportunity to collect an impressive amount of data on numerous animal species and fossils. Proceeding by inductive generalisation, Darwin developed a theory of evolution based on the idea of random and heritable variation, in which the action of natural selection intervenes in favour of the most suitable organism in a given environment. Darwin also made a proposal about the mechanism of inheritance of variation, called pangenesis, and formulated it in a variety of animals and plants under domestication (1868). According to this hypothesis, each part of the body produces corpuscles, the pangens, which flow into the egg cells and from there participate in the reproductive mechanism.

By contrast, Mendel based his theories on an extensive series of experiments conducted between 1856 and 1863 in Brünn, where he had joined the Augustinian monastery and taught as a substitute teacher in secondary schools. Based on crossbreeding generations of *Pisum sativum*, the common pea plant, Mendel studied the transmission and recombination of hereditary traits in offspring. In light of the crossbreeding obtained, he hypothesized the existence of two factors, one from the father and one from the mother, one of which could 'hide' the other. What is most significant about Mendel's methodology is not only the use of a quantitative and experimental approach but also, and above all, the application of mathematical and statistical calculations to the results obtained.

But why did Mendel's work go largely unnoticed? Several hypotheses have been proposed,[8] but none seems able to fully explain the substantial indifference of the scientific community of the time to the conclusions Mendel presented to the Natural Science Society in Brünn and published in its Proceedings. Whatever the reason, it was not until the beginning of the twentieth century that Hugo De Vries (1848–1935) and Carl Erich Correns (1864–1933) rediscovered the Czech monk's findings, which only then began to gain popularity and meet with experimental confirmation and general appreciation. At this point, a crucial figure in the history of genetics entered the scene: the Englishman William Bateson (1861–1926), the main advocate of Mendelian theory, who contributed greatly to its dissemination in English-speaking countries.

Through new studies, it became essential to investigate whether Mendelian factors were physical realities—and if so, where they were located—and how the transmission of a certain factor from par-

[6] A synthesis can be found in MAYR, *Storia del pensiero biologico* (ed. or. *The Growth of Biological Thought: Diversity, Evolution and Inheritance*, Cambridge, 1982), Torino, 2011, vol. I, pp. 589-599.

[7] J. VAN WYHE, *Mind the Gap: Did Darwin Avoid Publishing his Theory for Many Years?*, in *Notes and Records of the Royal Society of London*, 61 (2), 2007, pp. 177-205.

[8] See P. ROSSI, *op. cit.*, vol. II, tome II, pp. 924-928.

ents to offspring took place. The encounter between experiments in the field of agro-zootechnics, which had always looked at the transmission of traits, and research in the field of cytology, which enabled us to gain a better understanding of the structure and composition of cells, thus became important.

In fact, the existence of DNA, albeit called by different names, and of chromosomes had been known since the second half of the nineteenth century. It was the Swiss Friedrich Miescher who in 1869 isolated DNA during a series of experiments on leukocytes. Inside white blood cells, Miescher found a phosphorous-rich compound and called it nuclein, because he realised that it was in the cell nucleus. Almost twenty years later, Albrecht Kossel (1853–1927), Nobel Prize winner in 1910, showed that nuclein was composed of a protein part and a non-protein part (nucleic acid). Proceeding with the analysis of nucleic acids, he identified adenine, cytosine, guanine, thymine, and uracil. At the time, however, it was certainly not possible to hypothesise that DNA was involved in the mechanisms of the transmission of hereditary traits.

The same applies to chromosomes. They were discovered in 1878 by Walther Flemming (1843–1905), who also studied the process of cell division, called mitosis, and they were named by Heinrich Wilhelm Gottfried von Waldeyer Hartz (1836–1921). Their link to the transmission of hereditary traits became evident only years later. Incidentally, until 1955, the exact number of chromosomes in the human karyotype was not known; it became clear thanks to the work of Joe-Hin Tjio (1919–2001) and Albert Levan (1905–1988), both trained plant cytologists.

William A. Cannon (1870–1958) and Walter Sutton (1877–1916) were among the first to imagine that Mendelian factors were physically located on these threadlike structures. Their collaboration at the Zoological Station of Naples with Theodor Boveri (1862–1915) was essential because Boveri had already achieved important results. In 1902, Sutton, working on the chromosomes of *Brachystola magna* (grasshopper), realised that the number of chromosomes was halved during meiosis and that each spermatozoon or egg cell therefore received only one chromosome per pair. In his article 'The Chromosomes in Heredity', published in the *Biological Bulletin* in 1903,[9] Sutton demonstrated the substantial consistency of his results with Mendel's conclusions and suggested that chromosomes were the physical basis of Mendel's laws of heredity.

This was the beginning of intense research activity into chromosomes and the localisation of Mendelian factors, attracting the interest of many scholars and research groups. It is precisely in this crucial step, epistemological even more than experimental, that we identify the end of formal genetics and the beginning of what can be called chromosome genetics. Of the many advances made in the decades that followed, we should at least mention the work of Nettie M. Stevens (1861–1912), who identified the male Y chromosome. The discovery was published in the famous 1905 *Studies in spermatogenesis with especial reference to the accessory chromosome*[10] and followed the results of Erwin C. McClung (1870–1946), who had traced sex determination to the X chromosome alone.

[9] W. SUTTON, *The Chromosomes in Heredity*, in *Biological Bulletin*, 4 (5), 1903, pp. 231-251.

[10] N. M. STEVENS, *Studies in Spermatogenesis with Especial Reference to the "Accessory Chromosome"*, Washington, D.C., 1905, Vol I. *Cfr*. S. G. BRUSH, *Nettie M. Stevens and the Discovery of Sex Determination by Chromosomes*, in *Isis*, 69, 1978, pp. 162-172.

ISSN 2284-4503

A strong impetus for these new lines of research came also from Thomas H. Morgan (1866–1945), 1933 winner of the Nobel Prize for Medicine, and his research group, which concentrated their experiments on *Drosophila melanogaster* (fruit fly). Initially, Morgan was inclined towards an epigenetic theory. He was also sceptical of Sutton's studies and more prone to believe that the cytoplasm, and not the nucleus (and therefore the chromosomes), played a major role in heredity. After studying the white-eye mutation on *Drosophila* and analysing its transmission to offspring, Morgan completely revised his position. He argued that genes were physically located on chromosomes, and he explained that certain 'Mendelian factors' recombined during meiosis according to the theories of a Belgian cytologist, Frans Alfons Janssens (1865–1924), who in 1904 was the first to describe the crossing-over phenomenon. Janssens had noted how, during meiosis, members of chromosome pairs roll over each other, facilitating the exchange of strand segments. The more two factors usually associated (e.g. wings and eye colour) end up splitting, the further apart their positions on the chromosomes must be. This was the basis for the first mapping attempts by Morgan's group in the following years, in particular by Alfred H. Sturtevant (1891–1970).

The established variability of genes and the phenomena of recombination intensified the interest in mutations, which also led to a challenging methodological question: How can we systematically study a phenomenon that is in itself random and unpredictable? Attempts were made to induce mutations in the laboratory. One of the scientists who worked hardest on this problem came from Morgan's group: Hermann Joseph Muller (1890–1967), Nobel Prize winner in Medicine in 1946, who demonstrated how the use of massive doses of X-rays could induce mutations in egg and sperm cells.[11] Muller also claimed, unlike many of his colleagues, that the mutations induced by this method were the same as those that occur naturally.[12] It was only later that scientists began to use chemical mutagens.

Mutational studies are another fundamental line of research intertwined with the hypotheses about the genes' role in controlling and regulating metabolic reactions. One of the earliest examples predates Muller's findings: Between 1902 and 1908, Archibald Garrod (1857–1936) observed that certain metabolic diseases, such as alkaptonuria, albinism, cystinuria, and pentosuria, are transmitted according to Mendel's laws, therefore assuming that their cause was a mutation in a hereditary factor. They are in fact all diseases caused by the lack of a specific enzyme. Garrod's hypotheses became relevant again in the 1940s, when George Beadle (1903–1989) and Edward Tatum (1909–1975) devised the famous "one gene–one enzyme" formulation (which later became "one gene–one polypeptide chain", in which, as we know, a triplet of bases codes for an amino acid) and inferred that the two main functions of genes were self-replication and the production of enzymes.

In the 1930s, the analysis of human chromosomes[13], which had hitherto been little studied, was made possible by the development of cytogenetic techniques, paving the way for human genetics

[11] H. J. MULLER, *Artificial Transmutation of the Gene*, in *Science*, 66, 1927, pp. 84-87; ID., *The Problem of Genic Modification*, in *Proceedings of the 5th International Congress* 1, 1928, pp. 234-260.

[12] H. J. MULLER, *Artificial Transmutation of the Gene*, pp. 84-87.

[13] There is no room here to talk about the evolutionary synthesis, which saw Darwinian evolutionism and Mendelian theory merge into a study of populations that accounted for the most recent results in the field of genetics and the tools of statistics. John Haldane (1892-1964), Sewall Wright (1889-1988), and Ronald Fisher (1890-1962) are regarded as its initiators. For a historical reconstruction signed by one of the protagonists of the debate, see E. MAYR, *op. cit.*, vol. II.

and the intensification of discoveries of genetically based diseases. In the 1940s, it was the turn of thalassaemia, whose genetic origin was demonstrated by two Italian researchers, Ida Bianco (1917–2006) and her husband Ezio Silvestroni (1905–1990), and by James V. Neel (1915–2000), who went on to study sickle-cell anaemia. As is well known, it was Linus Pauling (1901–1994), later awarded the Nobel Prize for Chemistry and Peace, who in 1949 investigated the physical properties of haemoglobin in the case of anaemia and finally confirmed the link between genes and protein synthesis. In the following decade, attention expanded to diseases caused by chromosomal abnormalities. In 1959, for example, Jérôme Lejeune (1926–1994), counting on the valuable collaboration of Marthe Gautier (1925–), a specialist in the analysis of cell cultures (and using Tjio and Levan's cytological technique), observed that Down's syndrome was linked not to the absence of a chromosome but to the presence of an extra chromosome.

In the meantime, the so-called Phage Group, an informal collaborative network comprising, among others, the physicist Max Delbrück (1906–1981), Alfred D. Hershey (1908–1997), and Salvador Luria (1912–1991), tackled a new problem that had become central, namely, whether genetic material was carried by proteins or by DNA, both of which were present in the nucleus. The experiments were carried out using a bacteriophage whose phosphorus parts had been radioactively charged, and the results gathered evidence that it was DNA that contained the hereditary information. They found that the virus released not proteins but only DNA into the bacterium to "colonise" it. This discovery earned Delbrück, Hershey, and Luria the Nobel Prize for Medicine in 1969.[14]

Increasing research into the properties of deoxyribonucleic acid led to the discovery of its double-helix structure in 1953. This work, carried out by James Watson (1928–) and Francis Crick (1916–2004), would never have been possible without the studies of Maurice Wilkins (1916–2004) and Rosalind Franklin (1920–1958). At this point, it was necessary to explain not only the mechanisms of DNA self-replication but also how protein synthesis was controlled. The identification of messenger RNA was not long in coming. Finally, in a 1961 article published in *Nature*,[15] Crick, together with Leslie Barnett (1920–2002), Sydney Brenner (1927–2019), and Richard Watts-Tobin (1934–), set out to decipher the genetic code: the sequence of bases was to be read in triplets, since each triplet (or codon) encodes an amino acid.

In the years that followed, the steps forward were countless and decisive. It would be too complex to go through all the stages: for example, the 1977 discovery of the existence of introns and exons, and therefore of the presence of non-coding DNA; the first steps in biotechnology; and the localisation in 1983 of the first polymorphic marker linked to a genetic disease, that relating to Huntington's chorea, found on chromosome 4.[16]

The growth of increasingly precise and complex knowledge led to the Human Genome Project in the late 1980s. Given the considerable costs involved in the project, the first of its kind in the life sciences, there was much criticism and discussion. In fact, the complete sequencing of the human genome

[14] The reasons can be found at www.nobelprize.org/prizes/medicine/1969/summary/, last accessed 20 January 2021.

[15] F. CRICK, L. BARNETT, S. BRENNER, R. WATTS-TOBIN, *General Nature of the Genetic Code for Proteins*, in *Nature*, 192, 1961, pp. 1227-1232.

[16] J. F. GUSELLA ET AL., *A polymorphic DNA marker genetically linked to Huntington's disease*, in *Nature*, 306, 1983, pp. 234-238.

definitively opened up a new phase in the history of genetics, of which we are today only seeing the first effects, centred on the possibility of intervening directly on genes, for example, to prevent or cure diseases by modifying DNA.

3. Evolving concepts

This brief, non-exhaustive chronological reconstruction shows how the history of genetics is marked by the birth and sequence of new concepts, whose meanings constantly change, intertwining with philosophical, historical, sociological, and cultural issues. Obviously, the concept of the gene is central, but it is not the only one to deserve attention. Other concepts, such as heredity, 'race', eugenics, and epigenetics, have played and continue to play an important role, demonstrating once again how and to what extent research into DNA, the evolution of species, and the mechanisms of cellular reproduction have catalysed, in the past as well as today, reflections and debates in the world of research and in society in equal measure.

As many contributions have shown,[17] the concept of the gene has taken on profoundly different meanings in the history of genetics, in relation both to the periods when studies on heredity were carried out and to the epistemological approach of the researchers dealing with it over time.

The term *gene* was introduced[18] in 1908 by Wilhelm Johannsen (1857–1927), a Danish botanist, and soon replaced a series of words that had been used to that point, from Mendel's *Merkmal*—'character', 'factor'—to the *Anlage*—'unit'—of the Dutchman De Vries, and the *Unit-character* proposed by Bateson. The problem lay in the semantic confusion that these terms failed to resolve: they could refer either to the visible characteristic or to the corresponding hereditary basis. It was for this reason that Johannsen introduced the clear distinction between 'phenotype' and 'genotype'. The term *gene* recalled *pangene*, which Darwin and De Vries had used to refer to hereditary particles. This lexical change was crucial and brought with it questions not only of semantics but also of epistemology, science, and philosophy. The first problem emerging with renewed clarity was about the relationship between gene and characteristic, the chemical and biological mechanisms relating them, and, above all, the nature of the gene itself. If phenotypic characteristic was the empirically 'seen', how was the gene to be understood? Was it a material reality or, rather, a unit of calculation? The *formal approach*, the term used to refer to the pre-molecular or classical phase of the history of genetics,[19] did not concern itself with what a gene actually and chemically was. Johannsen himself was not interested in answering the question, as this passage from his fundamental text *Elemente der exakten Erblichkeitslehre*[20] shows: "The word gene is completely free from any hypothesis; it only ex-

[17] For an in-depth discussion: E.F. KELLER, *The Century of the Gene*, Cambridge, 2000; P. J. BEURTON, R. FALK, H.-J. RHEINBERGER, *The Concept of the Gene in Development and Evolution: Historical and Epistemological Perspectives,* Cambridge, 2000; P. PORTIN, *The Concept of the Gene: Short History and Present Status*, in *The Quarterly Review of Biology*, 68 (2), 1993, pp. 173-223; E. A. CARLSON, *Defining the Gene: An Evolving Concept*, in *American Journal of Human Genetics*, 49, 1991, pp. 475-487.

[18] Curiously, William Bateson had already used the term *genetics* in a private letter four years earlier, but it did not become widespread until after Johannsen introduced *gene*.

[19] P. PORTIN, *op. cit.*, pp. 175-179.

[20] W. JOHANNSEN, *Elemente der exakten Erblichkeitslehre*, Jena, 1909.

presses the established fact, that at least many properties of an organism are conditioned by special, separable, and thus independent 'conditions', 'foundations', 'dispositions'".[21] In the third edition of 1926, Johannsen emphasised the concrete dimension of the gene[22]—"Genes are realities, not hypothetical conceptions"—but then concluded that genes are "entities of calculation, expressions of realities of unknown nature".[23] Despite the possible apparent contradictions, it is quite clear that for Johannsen, it was important to state that genes exist, that "they are there". It was less important to establish "what they are" because that is not functional in explaining the mechanisms of heredity.

Already the cytogenetic phase implied a change since with Sutton and then the studies of Morgan and his research group, interest shifted to chromosomes,[24] their constitution, and the possible location of genes. Regarding this period, Hermann Muller's text, read at a conference in 1926, remains fundamental. The American geneticist reiterated his opinion that the gene was *the* basis of life and not just *a* basis of life. Incidentally, Muller argued for the key role of mutations and, more than anything else, their inheritance, maintaining that mutations and inheritance were key components of the same mechanism, not separate processes.[25]

A further breakthrough occurred with the entry into the molecular phase—which Portin calls *neo-classical*—and thus with the identification of the double-helix structure of DNA and the coding mechanisms of polypeptide chains. The years that followed 1953 strongly put the concept of the gene as a unit of information to the test. Consider, for example, the 1976 discovery of introns, demonstrating the existence of non-coding—and therefore apparently unused and unusable material—within each gene. Other examples include all the subsequent experiments that led to intervention directly on the gene to modify it. The more the gene was investigated, the more its concept became complex and difficult to handle. The words of Portin, who wrote thirty years ago, are very effective in this respect:

> [Due to] the discoveries of repeated genes, split genes, nested genes, overlapping genes, transposable genes, alter-native splicing, multiple and complex promoters, enhancers and silencers, downstream signals, internal control signals, proteolytic cleavage of translation products and other types of protein processing, editing of primary transcripts, the special case of immunoglobin genes, and the detailed analysis of gene complexes […], our comprehension of the nature of the gene entered a dramatic new phase. Paradoxically, in spite of the new, sometimes overwhelming, concreteness of our comprehension of the gene as a result of DNA technology, we seem to be left with a rather abstract and generalized concept of the gene that has quite different significances in different contexts.[26]

[21] Translation by N. ROLL-HANSEN, *Commentary: Wilhelm Johannsen and the problem of heredity at the turn of the 19th century*, in *International Journal of Epidemiology*, 43(4), 2014, pp. 1007-1013.
[22] W. JOHANNSEN, *Elemente der exakten Erblichkeitslehre,* Jena, 1926[3].
[23] N. ROLL-HANSEN, *op. cit.*, p. 1011.
[24] E. A. CARLSON, *The Drosophila Group: The Transition from the Mendelian Unit to the Individual Gene*, in *Journal of the History of Biology*, 7 (1), 1974, pp. 31-48.
[25] E. A. CARLSON, *The Drosophila Group: The Transition from the Mendelian Unit to the Individual Gene*, *op. cit.*, p. 31. See H. J. MULLER, *Variation Due to Change in the Individual Gene*, in *The American Naturalist*, 56 (642), 1922, pp. 32-50.
[26] P. PORTIN, *op. cit.*, p. 174.

It is precisely this last passage, in which Portin maintains that the concept of the gene "has quite different meanings in different contexts", that brings us back to the contemporary idea of the gene as a "boundary object" that crosses spheres and contexts and takes on renewed semantic values over time. In particular, we should emphasise how today the 'gene' is not an exclusive preserve of scientific language, belonging plastically to social, cultural, legal, political, and economic reality.

Moreover, it is typical of the history of genetics, as we said, to cross heterogeneous dimensions that go beyond the purely scientific. Another concept clearly demonstrates this: namely, the concept of eugenics.[27]

Well before the sad and well-known drifts of the Nazi and Fascist regimes, the term *eugenics* was coined in 1883 by Francis Galton (1822–1911),[28] who would later define it as "the study of the agencies under social control that may improve or impair the racial qualities of future generations either physically or mentally".[29] At the beginning of the twentieth century, eugenics programmes multiplied, together with the number of conferences, societies, and journals. Examples include the American Eugenics Record Office, the English Eugenics Society dedicated to popularisation, and the Eugenics Review.[30] The basic idea was to "improve" the human species by eliminating phenomena such as alcoholism, crime, 'moral degeneration', and even innate eroticism. The basic assumption of the programme was that all these behaviours were phenotypical traits determined solely and exclusively by a single gene (the environment or situations of degradation were not taken into account). It would therefore be easy to eliminate them, for example, by means of targeted sterilisation campaigns. Scientists of the calibre of Muller or Morgan adhered to this idea, and across the USA in the 1930s, a series of forced sterilisations were carried out. Opposition to the concept of eugenics became very strong in scientific circles, and fierce criticism arose because of factors including Hitler's rise to power, together with the Nazis' use of these theories, and the gradual awareness of the lack of scientific basis for the concept (it was soon discovered that each phenotypic characteristic was in fact determined by several genes). So, the term was no longer used, except in a negative sense, and the first human genetics programmes promoted at first the protection of the human gene pool and its variability as a guarantee of the survival of the species.[31]

A similar path and fate were met by the concept of race. The discoveries about the extreme variability of human DNA and the evolution of the human species demonstrated its scientific unreliability and definitively confirmed the impossibility of implementing a discrete and objective classification on a genetic basis.[32] The concept of race was so criticised by scientists that this criticism, combined with political events and social and cultural changes of the second half of the twentieth century, made necessary and imperative a broader reappraisal of the term.

[27] See A. Bashford, P. Levine (eds.), *The Oxford Handbook of the History of Eugenics*, Oxford, 2010, and also F. Cassata, *Molti, sani e forti. L'eugenetica in Italia*, Torino, 2006.

[28] F. Galton, *Inquiries into Human Faculty and its Development,* London, 1883.

[29] F. Galton, *Memories of my Life*, London, 1908, p. 321.

[30] L. Bland, L. A Hall, *Eugenics in Britain: The View from the Metropole*, in A. Bashford, P. Levine (eds.), *op. cit.*, pp. 213-227.

[31] B. Fantini, in P. Rossi, *op. cit.*, 1988, Vol. III, tome II, p. 793.

[32] L. L. Cavalli-Sforza, P. Menozzi, A. Piazza, *The History and Geography of Human Genes*, Princeton, 1994, pp. 16-20.

So these terms were decisively abandoned, but others were not. Let us mention at least one that was introduced in 1942 and continues to be used today. This is the concept of epigenetics, a recent branch of genetics studying how the environment influences gene expression without altering DNA sequences.

4. Conclusions: Contexts, actors, dynamics

The history of genetics is an incredible magnifying glass, allowing us to take a close look at science and how it works and has worked over the last century. If we look at the places and actors of gene studies, we find not only a sequence of hypotheses, discoveries, and methodologies but also a rich overview of contexts, places, and sociocultural dynamics.

Let us begin by looking at the two scientists who, as we said, are generally referred to as those who gave the decisive impetus to the research on evolution and the heredity of traits developed during the twentieth century: Mendel and Darwin. Their biographies and intellectual paths, in many different ways, open a window on what it meant to be a "man of science" in the second half of the nineteenth century. Mendel was the son of a farmer, and to continue his studies, he entered a monastery, where his appointment as abbot effectively prevented him from continuing his scientific work. Darwin, on the other hand, came from a wealthy family; the son of a doctor and grandson of a natural philosopher, he could support his travels and research without financial worries. For both of them, being a scientist was not a profession; it was an occupation developed according to their own inclinations and interests without any income. This point is crucial for the history of scientific thought as well as for the evolution of the figure of the scientist, especially in relationship with society.[33] Only during the twentieth century, due to the progressive institutionalisation of science and the production of knowledge, does the figure of the professional scientist emerge as one who researches no longer for (only) mere vocation but (also) for work, earning a living. This is a paradigmatic change, giving rise to new areas of knowledge and provoking new questions and reflections by social actors. It is therefore no coincidence that Craig Venter's (1946–) decision to found Celera Genomics as a private response to the Human Genome Project has catalysed growing debate and reflection on the lawfulness and morality of his conduct.[34] This scientist-entrepreneur personifies the highest point of this ongoing tension. On the one hand, there is the ancient perception of the scientist as a morally upstanding figure, who carries out his or her studies with complete economic disinterest. On the other hand, the professional scientist emerges, pursuing profit as a 'side effect' and using scientific research as a genuine entrepreneurial activity.

[33] On the images of the scientist throughout the history of science, see the fundamental works by Shapin, in particular S. SHAPIN, *The Way We Trust Now: The Authority of Science and the Character of the Scientist*, in P. HOODBHOY, D. GLASER, S. SHAPIN (eds.), *Trust Me, I'm a Scientist*, London, 2004, pp. 42-63; S. SHAPIN, *The Man of Science*, in L. DASTON, K. PARK (eds.), *The Cambridge History of Science. Vol. 3: Early Modern Science*, Cambridge, 2006, pp. 179-191; S. SHAPIN, *Figures de scientifiques*, in *Histoire des sciences et des savoirs, Vol. 3: Le siècle des technosciences (depuis 1914)*, Paris, 2015, pp. 27-45.

[34] For instance, S. SHAPIN, *Figures de scientifiques*, pp. 27-45; J. WITKOWSKI, *A life worth writing about*, in *Nature*, 449, 2007, pp. 785-786.

Furthermore, when the subject of the research is the genome, and therefore the very essence of human life, intense ethical, legal, and philosophical questions arise, summed up by this question: Who owns the genome?[35]

Another trend in the history of genetics relates to the birth of the laboratory in the modern sense. Consider Mendel and Darwin; surely, they were part of a network of contacts fundamental to their training and to the reception of their work. But it is clear that the two men of science worked on their research largely alone. We have to go forward in time to find more extensive, continuous, and even institutionalised collaborations and arrive at the first genuine research groups in the life sciences. One of the earliest, and also best known and most important, laboratories in the history of genetics is undoubtedly the one set up around the figure of Morgan, also known as the 'Drosophila Group', with reference to the animal model used, or the 'Fly Group', after the name of the room where the researchers met. Here, in the Zoology Department of Columbia University, biologists such as Muller, Sturtevant, and Calvin B. Bridges (1889–1938)[36] worked together. Another example is the famous Phage Group, an informal group that revolved around the figure of Delbrück rather than a real laboratory, in which the meeting of scientists from different backgrounds played a key role. This group gave rise to the Phage Course, a summer school at the Cold Spring Harbor Laboratory, which was fundamental to the training of numerous researchers.

It is even more interesting to note that, from the laboratory's limited and restricted size, the range of action has increasingly expanded, first with the intensification of collaborations between different research institutes, and then with the launch of the Human Genome Project,[37] a proper window on the Big Science that characterises our century and finds its counterpart in other fields in the Large Hadron Collider (LHC) of CERN in Geneva or in the International Space Station. The International Human Genome Sequencing Consortium has come to involve not only the USA and the UK but also Germany, Japan, China, and France.[38]

Finally, it should not be forgotten that the history of genetics, like the whole of the history of scientific knowledge, is also crowded with female figures that the historical memory has given us with difficulty despite the fundamental contributions they have made. The most famous is certainly Rosalind Franklin, but she is not the only one. Think of Nettie Maria Stevens, who discovered the Y chromosome; Barbara McClintock, winner of the Nobel Prize for Medicine and Physiology in 1983, who in the 1950s discovered transposons (genetic elements that can move); or Martha Chase, who performed the crucial experiment with bacteriophages that led to the identification of DNA as the carrier of genetic material: the experiment is now known as the Hershey-Chase experiment, yet Chase's collaboration with the Phage Group is rarely mentioned.

[35] M. ANGRIST, R. M. COOK-DEEGAN, *Who Owns the Genome?*, in *The New Atlantis*, 11, 2006, pp. 87-96.

[36] See E. A. CARLSON, *The Drosophila Group: The Transition from the Mendelian Unit to the Individual Gene*, *op. cit*. On the relevance of specific laboratory dynamics in the construction of scientific knowledge, see B. LATOUR, S. WOOLGAR, *Laboratory Life*: *The Construction of Scientific Facts*, Beverly Hills, 1979; B. LATOUR, *Science in Action: How to Follow Scientists and Engineers Through Society*, Cambridge, 1987.

[37] The bibliography is very rich. See, for example, H. ZWART, *Human Genome Project: History and Assessment*, in *International Encyclopedia of Social & Behavioral Sciences*, Oxford, 2015, 2 ed., pp. 311-317.

[38] www.genome.gov/human-genome-project. Last visited on 18 January 2021.

All these dynamics, which the history of genetics highlights, are a window on the broader panorama of the development of scientific thought. Genetics is undoubtedly a privileged place of observation. It tells us a great deal about our past and our present and will continue to do so for those who look back in the future.

ISSN 2284-4503

Special issue

The legacy and future of race between science, constitutional lexicon, and political action

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ABSTRACT: Beyond the recurring questions "Is race social or biological?", "Is race real or illusory?" which encapsulate the philosophical morass of the ontology of race, this paper, after a brief summary of the development of the scientific debate, focuses on the attitude that the law should take towards this contested concept and the "cursed" word that describes it. In particular, the analysis will focus on a "symbolic" aspect, that of the use of the word "race" in the constitutional lexicon of different countries, and on a more "functional" one, related to the use of the "racial argument" in some policies aimed at overcoming health disparities. Lastly, a brief reference will be made to the impact of technological innovations on the racial discourse, to understand, in particular, whether and how A.I. technologies increase or reduce racial bias.

KEYWORDS: Race; discrimination law; equality; genetics; constitutional law

SUMMARY: 1. A definitional puzzle: The multifaceted, illusory, "cursed" concept of race – 2. The origins and present-day relevance of race as a scientific assumption – 3. The two-fold attitude of the law to race – 3.1. Can constitutional amendments provide a solution? – 3.2. The contribution of race to (in)justice and (in)equality: From blindness to consciousness – 4. From yesterday's discrimination to tomorrow's: A.I. as a multiplier or reducer of racial bias?

1. A definitional puzzle: The multifaceted, illusory, "cursed" concept of race

The term race is common in scientific, social, political, and legal discourse around the world. The word, with its uncertain etymology,[1] refers to a multifaceted concept with a long history which has, over the years, developed a complex relationship with biology, social sciences, and the law. Its significance is due to its position at the interface between science and society. The concept, shaped by its surroundings over generations, has historically been used as a mechanism to group human beings and, consequently, as a powerful tool to legitimize systemic oppression.[2]

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[1] According to some authors, the term derived from "ratio", but others have provided evidence demonstrating that it originates from "haraz", an old French word which refers to horse breeding (G. FRANZINETTI, *The Historical Lexicon of Nationalism: Ethnicity, Ethnos, Race, Volksstamm: Historical Footnotes*, in *Colloquia Humanistica*, 5, 2016, p. 56).

[2] H. MOHSEN, *Race and Genetics: Somber History, Troubled Present*, in *Yale Journal of biology and medicine*, 93(1), 2020, pp. 215-219.

The concept's long use to justify the hierarchization of society links it explicitly with power, and some scholars have, in fact, conceived race and racisms (understood as the "political abuse of science"[3]) as structural and formative features of the colonialist era[4]. Claims of racial superiority — supposedly authorized by the laws of nature – have lurked in many places and times, producing aberrant episodes of human history, such as the Holocaust and apartheid, all interwoven from the threads of race and nature.[5]

The category of race has imbued social relations so deeply that Bauman, in his study of the Holocaust, identified the need for modernity to carefully construct a "new naturalness", grounded in an authority "different from that of the evidence of sensual impressions".[6]

Race can only be analyzed and understood within the interplay of politics and science. And now, genetics, which had historically provided an alleged basis for oppressive policies and the spread of systemic racism, is contributing to the delegitimization of "race". Scientific knowledge employed to biologize imaginary hierarchies of human variation has been superseded by the best research in genetics, revealing that the concept of "race" is elusive and has no scientific basis or reliable definition. The link with science does not, however, seem to have been completely severed, and the category of race has not yet altogether disappeared from the field of scientific research and medicine.

Beyond the recurring questions "Is race social or biological?", "Is race real or illusory?" which encapsulate the philosophical morass of the ontology of race, this paper, after a brief summary of the development of the scientific debate, focuses on the attitude that the law should take towards this contested concept and the "cursed"[7] word that describes it. In particular, the analysis will focus on a "symbolic" aspect, that of the use of the word "race" in the constitutional lexicon of different countries, and on a more "functional" one, related to the use of the "racial argument" in some policies aimed at overcoming health disparities. Lastly, a brief reference will be made to the impact of technological innovations on the racial discourse, to understand, in particular, whether and how A.I. technologies increase or reduce racial bias.

2. The origins and present-day relevance of race as a scientific assumption

While the origins of the term and its earliest uses are debated,[8] the best known race-based classification of human beings was proposed by the Swedish naturalist Carl Linnaeus who

[3] R. BENEDICT, M. MEAD, *Race: science and politics*, Athens, 2019, p. vii.

[4] A. STOLER, *Race and the Education of Desire: Foucault's History of Sexuality and the Colonial Order of Things*, Durham, 1995, p. 9.

[5] See D. S. MOORE, J. KOSEK, A. PANDIAN (eds.), *Race, nature, and the politics of difference*, Durham and London, 2003, Introduction, pp. 1-70.

[6] Z. BAUMAN, *Modernity and the Holocaust*, Cambridge, 1989, p. 57.

[7] This definition was given in Italy during the debates of the Constitutional assembly by Meuccio Ruini (see the Constitutional Assembly's debates of 24 March 1947 in www.nascitacostituzione.it).

[8] It is interesting to note that, according to an analysis of the uses of the word "race" in different ages, the physical-biological connotation of the term does not appear until the 18th century. See D. TEYSSEIRE, *De l'usage historico-politique de race entre 1680 et 1820 et de sa transformation*, in *Mots. Les langages du politique*, 33, 1992, pp. 43-52 and N. HUDSON, *From "Nation" to "Race": The Origin of Racial Classification in Eighteenth-Century Thought*, in *Eighteenth-Century Studies*, 29(3), 1996, pp. 247-264.

classified *Homo sapiens* into four groups: *europaeus*, *americanus*, *asiaticus*, and *afer*. By the tenth edition of his *Systema Naturae*, published in 1758, these groups had become subspecies, color-coded as red Americans, white Europeans, yellow Asians and black Africans.[9] At the time, various attempts were made to categorize race scientifically, influenced by Linnaeus' inclusion of social and personality traits alongside physical ones in order to further entrench racial hierarchies.[10]

The more cautious approaches developed by Darwin, who challenged the rigidity of these categories and emphasized the inconsistency and subjectivity of the criteria chosen to define them[11], were followed by the ideas of Galton, who theorized eugenics as the science by which the human stock would be improved and described how selective breeding could be used to give "more suitable races [...] a better chance of prevailing".[12] Galton's ideas, shared by a considerable number of other members of the scientific establishment at the time, spread and became increasingly influential during the second half of the 1800s[13].

Early twentieth-century manifestations of the concept marked an important change: whereas previous theories had based racial distinctions on observable physical traits, such as cranial capacity and skin color, as knowledge of biology advanced, race came to be conceived as a reflection of unseen differences attributed to the then newly discovered rules of heredity, based on genes. Eugenics progressively gained momentum, justifying practices of discrimination and marginalization in a range of different ways.

It took the Nazi policies of "racial hygiene" and the Holocaust to show the world the logical endpoint of this horrific ideology[14]. And, while these theories are no longer part of the scientific debate, and openly eugenic practices are no longer tolerated,[15] the confused relationship between race and biology has persisted.

1972 marked a turning point in the debate on race, when Harvard geneticist Richard Lewontin, in a paper titled "The Apportionment of Human Diversity," showed that most genetic variation (85.4 %) occurs within human populations, and not among (supposed) racial groups. Lewontin concluded that

[9] S. MÜLLER-WILLE, *Linnaeus and the Four Corners of the World*, in K.A. COLES, R. BAUER, Z. NUNES, C.L. PETERSON (eds), *The Cultural Politics of Blood, 1500–1900*, London, 2014, pp. 191-209.

[10] H. MOHSEN, *Race and Genetics: Somber History, Troubled Present*.

[11] C. DARWIN in his *On the Origins of Species* (1859) and *The Descent of Man, and Selection in Relation to Sex* (1871). For a reconstruction of the evolution of the concept, see P. Greco, *Breve storia del concetto di razza umana*, in M. Monti, C.A. REDI (ed.), *No razza, sì cittadinanza*, Pavia, 2017, pp. 21-29 and Id., *Addio alla razza. Una parola pericolosa che per la scienza non ha senso*, in *Scienza e Società*, 27-28, pp. XIII-133.

[12] F. GALTON, *Inquiries into Human Faculty*, London, 1883, p. 25.

[13] On the historical evolution of genetics, in this Special Issue, see I. AMPOLLINI, *A brief history of genetics: chronology, concepts, themes*.

[14] A fundamental reference goes to Gobineau's *Essay on the inequality of races*, which had an almost incalculable effect on late 19th-century social theory. Published in 1853-55, it is thought to be the "intellectual" link between earlier racial theories and later, explicitly discriminatory scientific systematizations and public policies (J.A. DE GOBINEAU, *Essai sur l'inégalité des races humaines*, Paris, 1833-1835).

[15] An intuition of race as a social concept with no genetic basis seems first to have been presented by the anthropologist Ashley MONTAGU in 1942, in his *Man's Most Dangerous Myth: The Fallacy of Race*, New York, 1942.

"Since such racial classification is now seen to be of virtually no genetic or taxonomic significance either, no justification can be offered for its continuance".[16]

The debate around the concept of race resurfaced with the conclusion of the Human Genome Project in 2003, which seemed to offer an important opportunity to finally lay the race and genetics debate to rest: announcing the working draft of the human genome, both Francis Collins and Craig Venter clearly declared the inexistence of a genetic basis for race. Only two years later, however, just before the official completion of the Project, a report was published in Science, which appeared, to some, to confirm the biological basis of race. The paper claimed that genetic samples from 52 populations could be clustered into five geographically based categories: Europe, Africa, East Asia, Oceania, and the Americas.[17] Then, in March 2003, an article in the *New England Journal of Medicine* argued, to the contrary, that "although everyone, from geneticists to laypersons, tends to use 'race' as if it were a scientific category, with rare exceptions, no one offers a quantifiable definition of what a race is in genetic terms."[18]

A decade and a half later, in 2018, the debate flared up again, ignited by the publication of an opinion piece in the *New York Times* titled "How Genetics is Changing our Understanding of 'Race'". Harvard geneticist David Reich argued that the consensus around Lewontin's positions (most of which the author shared) had calcified over the decades, into an orthodoxy. In particular, Reich contested the wide-spread anxiety about any research into genetic differences among populations, supposed to be poised "on a slippery slope" that can lead to the kinds of pseudoscientific arguments that were used to try to legitimize the slave trade, the eugenics movement and the Nazis' murder of six million Jews. Reich expressed his "deep sympathy for the concern that genetic discoveries could be misused to justify racism", but said that nevertheless, as a geneticist, he was also clear "that it is simply no longer possible to ignore average genetic differences among "races." Reich's concern was that advances in the field of genetics could reveal substantial biological differences among human populations, and that those discoveries might then be cited as "scientific proof" that racist prejudices and agendas are correct. The topic's sensitivity was immediately demonstrated by a response to Reich's opinion, given by 67 scholars (from disciplines ranging across the natural, medical and population health and social sciences to law and the humanities) in an open letter, titled "How Not to Talk About Race and Genetics". The letter's intention was to highlight the complexity of the concept of race and the fact that it does not operate in a political vacuum. Relying on a robust body of scholarship recognizing the existence of geographically based genetic variation in our species, but at the same time showing that such variation is not consistent with biological definitions of race, the letter called for a "a more sophisticated approach to the problem of human group categorization in the biomedical sciences."

[16] R.C. LEWONTIN, *The Apportionment of Human Diversity*, in T. DOBZHANSKY, M.K. HECHT, W.C. STEERE (eds), *Evolutionary Biology*, New York, 1972.

[17] N.A. ROSENBERG et al., *Genetic Structure of Human Populations,* in Science, 298(5602), 2002, pp. 2381-2385. See also E.D. LANDER, L.M. LINTON, B. BIRREN et al., *Initial sequencing and analysis of the human genome*, in *Nature*, 409, 2001, pp. 860-921.

[18] R.S. COOPER et al., *Race and Genomics*, in *The New England Journal of Medicine*, 348, 2003, pp. 1166-1170.

While the general consensus among Western researchers is now that human race is a sociocultural construct, and not a genetic variable,[19] references to "race" or "racial categories" still occur in biomedical research and medical practice, revealing the persistence of biologic essentialism.[20]
One of the reasons why racial labels are still in use is that a great deal of research is currently being done into human biological differences, in order to develop "precision medicine" instruments, diagnostic and therapeutic tools that are as well suited as possible to individuals' genetic makeup.[21]
Many, therefore, consider the concept of race to provide a useful, if imperfect, method for grouping people. Researchers often rely on self-reported race to assess people's origins,[22] despite the fact that this criterion has no scientific basis and does not correspond to real variability, especially in case of admixed populations. Furthermore, the applicability of data drawn from these studies is often weakened by researchers' failure to explain what they mean by "race," as if its meaning is self-evident.[23]

[19] In a very clear synthesis, the American Association of Physical Anthropologists states that "Race does not provide an accurate representation of human biological variation. It was never accurate in the past, and it remains inaccurate when referencing contemporary human populations. Humans are not divided biologically into distinct continental types or racial genetic clusters. Instead, the Western concept of race must be understood as a classification system that emerged from, and in support of, European colonialism, oppression, and discrimination. It thus does not have its roots in biological reality, but in policies of discrimination. Because of that, over the last five centuries, race has become a social reality that structures societies and how we experience the world. In this regard, race is real, as is racism, and both have real biological consequences" (*Executive Summary: AAPA Statement on Race and Racism*, March 8, 2019).

[20] R.S. COOPER, *Race in Biological and Biomedical Research*, in *Cold Spring Harbor Perspectives in Medicine*, 3(11), 2013. See also V. L. BONHAM, E.D.GREEN, J.PEREZ-STABLE, *Examining how race, ethnicity, and ancestry data are used in biomedical research*, in *JAMA*, 320(15), 2018, pp. 1533-1534.

[21] So-called "personalized/precision medicine" refers to the tailoring of medical treatment to the individual characteristics of each patient. "It does not literally mean the creation of drugs or medical devices that are unique to a patient, but rather the ability to classify individuals into subpopulations that differ in their susceptibility to a particular disease, in the biology and/or prognosis of those diseases they may develop, or in their response to a specific treatment" (NATIONAL RESEARCH COUNCIL, Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease, Washington D.C., 2011, p. 125). Nevertheless, the use of the notion of race for these purposes is widely contested: see G. ADIGBILI, *Race, science and (im)precision medicine*, in *Nature*. Medicine, 26, 2020, pp. 1675-1676.

[22] This criterion is strongly supported by some authors who consider self-identified race as a useful factor for understanding differences in disease and in the response to drugs: see among others, N. RISCH, E. BURCHARD, E. ZIV, H. TANG, *Categorization of Humans in Biomedical Research: genes, race and disease*, in *Genome Biology*, 3(7), 2002, pp. 1-12, according to which "self-defined race, ethnicity or ancestry are actually more genetically informative than clusters based on analysis of random genetic markers" (p. 6). Others highlight how "Group identity (…) and genetic heritage are much more complex than self-identity" (T.B. MERSHA, T. ABEBE, *Self-reported race/ethnicity in the age of genomic research: its potential impact on understanding health disparities*, in *Human Genomics*, 9, 2015, pp. 1-15.

[23] One paper examined how race and ethnicity terms are used in publications on genetic research and, sampling over 300 studies published between 2001 and 2004, found that fewer than 10 percent of the articles contained any discussion of how racial labels were assigned (P. SANKAR, M.K. CHO, J. MOUNTAIN, *Race and ethnicity in genetic research*, in American Journal of Medical Genetics, Part A, 143A, 2007, pp. 961–970). Furthermore, it has to be acknowledged that racial categories are dynamic designations which have to be understood as specific to the time and place in which they occur and are constantly being redefined. For example, in the U.S. census racial categories have changed every decade since 1790 (M. NOBLES, *Shades of citizenship: Race and the census in modern politics*, Stanford, 2000).

In addition to being inaccurate, the use of an unspecified criterion of race can be dangerous, for at least two reasons: on the one hand it can reinforce the assumption that human races have a biological basis and, on the other, it can blind scientists to other factors that contribute much more to health disparities.

Despite frequent attempts to replace "race" with other terms, free of stigma or oriented to show how humanity can be organized but not hierarchized (such as "ethnicity" or "ancestry") and despite mounting scientific evidence showing that humans are fundamentally more similar than different from each other, the legacy of the past still shadows genomic research. For example, even though ancestry testing is often marketed as a celebration of diversity and a way to overcome difference, racial supremacists, such as members of the Alt-right in the U.S., have embraced these tests as a way to prove their "pure white heritage" (while often rejecting or reinterpreting "unsatisfactory" results).[24]

Regardless of the words chosen, the concept of race is an epistemological category that operates independently of the aims of any individual geneticist. That said, the latter should not ignore the social implications of what they do.[25]

3. The two-fold attitude of the law to race

Reference to race in the legal sphere can be interrogated from two perspectives. First, in view of the "expressive"[26] and therefore also symbolic value of the law, should the use of the term in legal - and particularly in constitutional - texts be reconsidered, or is it appropriate to retain it?

Secondly, it is necessary to acknowledge the perennial ambivalence of the concept of race in the legal domain. On the one hand, especially in the past, public power seized upon it, using "race" as a pseudo-scientific argument for the categorization and hierarchization of human beings and thereby causing tangible harms. Conversely, more recently, "race" has been employed by the law to protect and benefit minority groups. It has also increasingly become an important identity factor, whether individually or collectively chosen.[27] The same concept has thus sometimes proved profoundly dangerous, at others, potentially empowering.

3.1. Can constitutional amendments provide a solution?

As the scientific debate has progressed, in recent years, so too has the law, which has also questioned the validity of the notion of race and, in particular, whether the term should enter, or remain in, the constitutional lexicons of various states. As is well known, the constitutions of many countries refer to the category of race, generally included as one of the factors that do not justify differential treatment.

[24] A. PANOFSKY, J. DONOVAN, *Genetic ancestry testing among white nationalists: From identity repair to citizen science*, in Social studies of science, 49(5), 2019, pp. 653-681. On the use of ancestry testing in different legal contexts, T. JONES, J.L. ROBERTS, *Genetic race? DNA, Ancestry tests, racial identity, and the law*, in *Columbia Law Review*, 120(7), 2020, pp. 1929-2016.

[25] R.S. COOPER et al., *Race and Genomics*, cit., p. 1169.

[26] On the "expressive" role of the law in shaping public ideology: C.R. SUNSTEIN, *On the Expressive Function of Law,* in *University of Pennsylvania Law Review*, 144, 1996, pp. 2021-2053.

[27] See the political ontology of race constructed by M.R. JAMES, *The political ontology of race*, in *Polity*, 44(1), 2012, pp. 122 ff.

ISSN 2284-4503

The word "race" was first used in legislation in 1870, when the United States decided to prohibit discrimination "on the basis of race" in the 15th Amendment to the Constitution.

In Europe, "race" first appeared in 1920 in the Treaty of St. Germain (which established peace between Austria and the Allied Powers), soon afterwards entering the Austrian and Czech Constitutions. After World War II, of course, "race" was heavily identified with Nazi ideology and thus considered a well-nigh pathogenic concept. It was within this context that it entered many post-War constitutions. Similar provisions can also be found in more than 140 more recent constitutions worldwide, such as those that emerged from the collapse of the Soviet bloc and apartheid South Africa, for instance. It is worthy of note that while fewer than half of the Constitutions adopted before 1970 include the protection of equal rights on the basis of race/ethnicity, 89% of those adopted between 2000 and 2009 and 79% of those adopted between 2010 and 2017 do so.[28]

The 1947 peace treaties obliged the defeated countries to prepare the necessary constitutional instruments to ensure the protection of fundamental rights "without distinction of race, sex, language or religion".[29]

Despite these international indications, and at a time when many people still believed in the scientific basis of human races, the problematic nature of the concept sometimes emerged during the drafting of constitutional texts. In Italy, for example, while art. 7 of the draft Constitution (which would later become art. 3) was being discussed, an amendment by Mario Cingolani was debated: "In the first paragraph, replace the word race with the word lineage (stirpe)". Cingolani justified his proposal by describing the need expressed in this regard by the Jewish communities, a need which deserved to be heard, in recognition of their recovery of absolute equality as Italian citizens.[30]

This proposal was opposed by other members of the Constituent Assembly, who considered that the term "race" (razza) should be maintained as it referred to historical fact, to acts of real discrimination which had taken place in Italy; the word was free of any negative connotation and its inclusion was intended to draw a line under what had happened in the past and to affirm equality among all human beings (Laconi and Ruini).

In Italy, however, the issue has recently re-emerged, in response to the consolidation of new scientific knowledge: on 14 October 2014, two scientists, Olga Rickards and Gianfranco Biondi, appealed to the President of the Republic, the Presidents of the Chambers of the Parliament and the President of the Council of Ministers to remove the term "race" from the Constitutional Charter, for lack of any objective basis. The ensuing, and ongoing, debate[31] has still not led to any concrete results.

[28] A significant exception is Latin America: the word "race" is not present in the constitutions of Argentina, Costa Rica, Guatemala, Haiti, Paraguay, the Dominican Republic or Uruguay. Interestingly, while Bolivia and Ecuador removed the word during recent constitutional reforms, it has made its way into the new constitutional text proposed in Chile (see www.constituteproject.org).

[29] See, for example, Art. 15 of the Treaty of peace with Italy (Paris, 10 February 1947), according to which "Italy shall take all measures necessary to secure to all persons under Italian jurisdiction, without distinction as to race, sex, language or religion, the enjoyment of human rights and of the fundamental freedoms, including freedom of expression, of press and publication, of religious worship, of political opinion and of public meeting."

[30] Constitutional Assembly debates of 24 March 1947 in www.nascitacostituzione.it.

[31] On the use of the word "race" in the Italian Constitution, among others, G. SILVESTRI, *Il termine "razza" nella Costituzione*, in A. MENICONI, M. PEZZETTI (eds.), Razza e inGiustizia, *Consiglio Superiore della Magistratura - Consiglio Nazionale Forense*, 2018, pp. 69-76; G.M. FLICK, *Verso la convivenza: leggi razziali, eguaglianza e pari*

In other jurisdictions, in contrast, more significant developments are occurring with regard to such references (now shown to be scientifically unfounded) and whether or not they should be retained.[32]
France is one of the countries where this issue has been most hotly debated. After the Second World War, France, like most European countries, banned racial discrimination in its 1946 Constitution, intended as a clear rejection of racist theses and 19th century theories of race. The use of the term "race" was not discussed, and it was also incorporated into the 1958 Constitution.
Then, in 1992, a scholarly debate around "race" was initiated by two law professors, Danièle Lochak and Jean-Jacques Israël, who were convinced that, since genetics had shown the term to have no basis in science, it should be abandoned. The proposal was that any legal text containing the word should be amended, replacing it with a reference to racism.
Almost ten years later, the Communist MP Michel Vaxès proposed removing the term "race" from the Constitution and other legislation,[33] but was opposed, mainly by those who feared that the removal of the term could mean the loss of a valuable tool for combating racism. Debating a similar proposal, advanced six years later, Jean-Luc Mélenchon suggested adding the adjective "prétendue" before the term "race", but this was also rejected. The debate did not entirely die down and was then rekindled a few years later, by a statement made by the then presidential candidate François Hollande: "Il n'y a pas de place dans la République pour la race. Et c'est pourquoi je demanderai, au lendemain de la présidentielle, au Parlement de supprimer le mot race de notre Constitution".
After another failed attempt to reform the French legislation,[34] the first tangible results were obtained when the Law for the modernization of justice in the 21st century[35] and the law on equality and citizenship[36] replaced the word "race" with the expression "so-called race" in some articles of the

dignità sociale, in *Rivista AIC*, 1, 2019, pp. 72-82; A. MORELLI, *Should Italy follow Germany's lead and remove 'race' from the Constitution?*, in *Il Manifesto Global Edition*, 6 December 2020, online: https://global.ilmanifesto.it/; V. TONDI DELLA MURA, *La parola razza nella Costituzione, ovvero: della rilevanza costituzionale di una nozione scientificamente infondata*, in *Diritti fondamentali*, 2, 2019, pp. 1-17; S. SALARDI, *"Razza": falsi miti e danni reali di un concetto. Abolirlo serve alla causa anti-discriminatoria?*, in *Materiali per una storia della cultura giuridica*, 2, 2016, pp. 451-478; V. SATTA, *Principio di eguaglianza e razza, tra dibattito costituente e giurisprudenza costituzionale*, in *Jus-online*, 5, 2020, pp. 298-330.

[32] Some intermediate solutions have been adopted in the course of constitutional revision processes. For instance, in Sweden, the 1974 Constitution was amended in 2012 and the term race, which was present in art. 15, prohibiting discrimination on various grounds (Art. 15. "No act of law or other provision may imply the unfavourable treatment of a citizen because he belongs to a minority group by reason of race, colour, or ethnic origin"), and also in articles 14 and 22.7, was cancelled and can now be found only in art. 4.11, among the limits to freedom of the press.

[33] Proposition of law n. 623: Proposition de loi *tendant à la* suppression *du mot* « race » *de notre* legislation, presented on 13 February 2003, online: https://www.assemblee-nationale.fr/12/propositions/pion0623.asp.

[34] In 2003 the National Assembly passed a bill to remove the word "race" from all French statutes (Proposition of law n. 218: Proposition de loi tendant à la suppression du mot « race » de notre législation, presented on 13 February 2003, online: https://www.assemblee-nationale.fr/14/pdf/propositions/pion0218.pdf).

[35] Loi n°2016-1547 du 18 novembre 2016 - Art. 86

[36] Loi n°2017-86 du 27 janvier 2017, Art. 171 and Art. 13.

criminal code.[37] Despite these and other changes, the term remains in some parts of French legislation.[38]

It also, moreover, remained in the Constitution. Nevertheless, in 2018 a proposal to remove "race" from art. 1 of the Constitution was advanced by LREM MPs,[39] using the now standard argument that the use of the term implied its reality. The bill was passed unanimously on Thursday 12 July 2018 by the Assemblée Nationale. However, the constitution has not been amended, as the process was interrupted by the so-called "Benalla affaire".

It is interesting to note, however, that even if the reform had taken place, "race" would have remained in the French constitutional lexicon, since the word also occurs in the preamble to the 1946[40] Constitution, having been included in the *bloc de constitutionnalité*.[41] No proposal to amend the latter has ever been made, since it is a text of historical value,[42] unlike the 1958 Constitution, which has been amended more than twenty times.[43]

The different value attributed to the two (1946, 1958) texts somewhat reflects the two main positions taken on these legal debates, which we will now explore in more detail: some believe that the word race has a symbolic, monitory value, and should therefore be maintained, particularly in constitutional texts, given their foundational nature, others believe that constitutional texts are living instruments which should be adapted to contemporary needs and facts.

In Germany too, and the history of the country's legal system lends this proposal particular significance, a large political majority (CDU-CSU-SPD) has recently signaled its intention to amend Article 3, Section 3 of the German Basic Law, according to which "No person shall be favoured or disfavoured because of his sex, parentage, race, language, homeland and origin, faith or religious or political opinions. No person shall be disfavored because of disability".[44]

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[37] See also Art. 225-1 and Articles 132-76 and 222-13 of the Code pénal.

[38] See, for example, Art. 695-9-17 of the Code de procédure pénale and Art. D4122-8 of the Code de la défense. Furthermore, the Code pénal still contains a reference to "groupe racial" (see. Art. 212-1).

[39] "Le terme de « race » a été introduit dans la Constitution en 1946, après le nazisme, pour indiquer que toute race était égale, pour mettre fin aux discriminations et rejeter les théories racistes. Toutefois, la persistance de sa mention est aujourd'hui mal comprise, à rebours de l'intention initiale. Il est donc proposé de supprimer ce terme de l'article premier" (Amendment CL847, adopted on 27 June 2018).

[40] See par. 1 ("[…] the people of France proclaim anew that each human being, without distinction of race, religion or creed, possesses sacred and inalienable rights") and par. 16 ("France shall form with its overseas peoples a Union founded upon equal rights and duties, without distinction of race or religion").

[41] As is well known, the *bloc de constitutionnalité* was "created" by the *Conseil Constitutionel* in a decision of 1971 (décision n° 71-44 DC) to compensate for the lack of a bill of rights in the 1958 Constitution. Relying on the preamble of the 1958 Constitution, which refers to the Preamble of the 1946 Constitution and the Declaration of 1789, the *Conseil* gave fundamental rights full constitutional value, allowing the *Conseil* to protect those rights and asserting itself as a modern constitutional court.

[42] Although nothing prevents a modification of these pieces of legislation, there is a broad scholarly consensus that the texts of 1789 and 1946 should not be modified or updated. On this, see the report of 2008 *Redécouvrir le Préambule de la Constitution - Rapport du comité présidé par Simone Veil*, online: https://www.vie-publique.fr/sites/default/files/rapport/pdf/084000758.pdf.

[43] The list of the constitutional amendments is available at: https://www.conseil-constitutionnel.fr/la-constitution/les-revisions-constitutionnelles.

[44] German Basic Law, 8 May 1948. In German: "Niemand darf wegen seines Geschlechtes, seiner Abstammung, seiner Rasse, seiner Sprache, seiner Heimat und Herkunft, seines Glaubens, seiner religiösen oder politischen

The current provision has been criticized on the grounds that the use of the word "race" could lead to the erroneous conclusion that the text recognizes the existence of categorical differences between human beings, a conclusion reinforced by the presence of the adjective "his" before the word "race". According to the German Institute for Human Rights (Deutsches Institut für Menschenrechte), the current wording of the article risks perpetuating racist thinking[45] and contains an irresolvable contradiction, requiring those affected by racial discrimination to declare that they have been discriminated against on the basis of their "race", thus making the use of racist terminology unavoidable.[46] According to others, however, the decisive argument would derive from the Basic Law's "negative" use of the term, since it rejects discrimination based on race, thereby stigmatizing both the word and the terminology related to it. The constitutional statement is thus connected with a historical tradition to which it is proposed as an alternative.[47]

The proposal put forward by the Federal Ministry of Justice and Consumer Protection, sent for comment to all bodies with an interest in the topic at the beginning of February 2021, charts a course between the two extremes: on the one hand, it suggests eliminating the direct reference to the word race, on the other - in order not to weaken an important barrier to the spread of racism - it proposes the addition of a reference to "racial motives". Specifically, the proposal is to delete the expression "seiner Rasse" and to add "oder aus rassistischen Gründen" at the end of the sentence. The new version of the article would thus read: "No person shall be favoured or disfavoured because of sex, parentage, language, homeland and origin, faith or religious or political opinions or on racist grounds". This wording is inspired by that already contained in the constitutions of some Länder, such as Brandenburg (art. 12, par. 2) and Sachsen-Anhalt (art. 7, par. 3).

The new wording, according to its proponents, would have the following advantages: the use of the adjective "racist" denotes an action or an opinion and would enable – even more explicitly than the contested notion of "race" - the inclusion of both actual and merely presumed biological features. Secondly, the abandonment of the term race means that the article transcends the origins of the formulation (a response to National Socialism), thus extending to cover wider forms of discrimination. Finally, the discussion paper clarifies that the use of the term "Gründen" rather than, for instance, "Kriterien", means that no objective connecting factors for discrimination are required.[48]

Anschauungen benachteiligt oder bevorzugt werden. Niemand darf wegen seiner Behinderung benachteiligt werden".

[45] H. CREMER, *Das Verbot rassistischer Diskriminierung. Vorschlag für eine Änderung von Artikel 3 Absatz 3*, Deutsches Institut für Menschenrechte, 2020, online: https://www.institut-fuer-menschenrechte.de/publikationen/detail/das-verbot-rassistischer-diskriminierung.

[46] H. CREMER, *Ein Grundgesetz ohne „Rasse" Vorschlag für eine Änderung von Artikel 3 Grundgesetz*, Deutsches Institut für Menschenrechte, 2020, online: https://www.institut-fuer-menschenrechte.de/fileadmin/_migrated/tx_commerce/policy_paper_16_ein_grundgesetz_ohne_rasse_01.pdf.

[47] See the debate between Hendrik Cremer (a researcher at the German Institute for Human Rights)and Uwe Volkmann (Professor of Public Law and Philosophy of Law at the University of Frankfurt), available at https://www.deutschlandfunk.de/hendrik-cremer-vs-uwe-volkmann-rasse-raus-eine-grundgesetz.2927.de.html?dram:article_id=478960.

[48] The new proposal and the reasons supporting it can be found in the draft Discussion at: https://www.bmjv.de/SharedDocs/Gesetzgebungsverfahren/Dokumente/DiskE_Ersetzung_Begriff_Rasse.pdf?__blob=publicationFile&v=3.

The proposed formula, however, has not escaped criticism since, according to some (e.g. the Greens), it would require discriminatory intent, thus privileging a subjective element rather than the objective circumstances, resulting in diminished levels of protection.

The Greens, in fact, had already submitted a different proposal, in which they suggested the deletion of the word race and the introduction of the adverb *rassistisch*. *Rassistisch Benachteiligung* (racial disadvantage) and *rassistisch Bevorzugung* (racial preference) should be understood objectively, like the other (substantive) prohibitions of discrimination of Article 3, section 3 of the Basic Law, and do not presuppose a subjective-intentional element.[49]

While Germany seems – at least according to some – to be ready to break with the past, a similar path may not be possible in legal systems where the scars of racism are even farther from being healed. The extent to which a system's history affects not only the structure and content but also the vocabulary of the constitutional text in the country concerned is evident from reading the 1996 South African Constitution, where the words "race", "racial" and "non-racialism" are mentioned in several provisions.

From a South African legal perspective, the European debate can appear "perplexing", since the removal of the term race from the Constitution could be read as a "regressive step aimed at protecting white privilege and reinforcing the social and economic dominance of the white minority". Here, the explicit and recurring references to race have a dual function, to "prevent the perpetuation of public and private forms of racial discrimination and racism" and "to address the effects of past and ongoing racial discrimination and racism by allowing or mandating race-based redress measures to correct the racial injustices of the past".[50]

The importance of the issue is clearly evident in Section 1 of the Constitution which lists "non-racialism" as one of the fundamental values of the whole constitutional system. Among the different interpretations of this principle, one is especially significant, particularly for the discourse being developed in this article. The South African Constitutional Court sees "non-racialism" as an ideal towards which to strive; the creation of a non-racial society has to start from an acceptance that "race" produces significant effects in daily life, otherwise the results of lingering racism and formal and informal racial discrimination cannot be dealt with effectively.[51]

This approach focuses on the function of the – albeit objectively unfounded – concept of race and the concrete effects it has in society.

[49] A further proposal is to add a sentence about the role of the State in protecting against any group-related violation of the equal dignity of all people and in working towards the elimination of existing disadvantages (Der Staat gewährleistet Schutz gegen jedwede gruppenbezogene Verletzung der gleichen Würde aller Menschen und wirkt auf die Beseitigung bestehender Nachteile hin)." The text of the proposal can be found at https://dserver.bundestag.de/btd/19/244/1924434.pdf.

[50] P. DE VOS, *"Race" and the Constitution: A South African perspective*, in *Verfasssungsblog.de*, 26 June 2020, at: https://verfassungsblog.de/race-and-the-constitution-a-south-african-perspective/.

[51] This was clearly stated by the Constitutional Court in a 2004 judgement: "However, it is also clear that the long-term goal of our society is a non-racial, non-sexist society in which each person will be recognised and treated as a human being of equal worth and dignity. Central to this vision is the recognition that ours is a diverse society, comprised of people of different races, different language groups, different religions and both sexes. This diversity, and our equality as citizens within it, is something our Constitution celebrates and protects." (*Minister of Finance and Other v. Van Heerden*, CCT 63/03 [2004], par. 44).

With reference to the constitutional language, in conclusion, it can be observed that the choice either to eliminate or to maintain the term race is profoundly influenced by each legal order's relationship with history, the substance of the social issues with which it is confronted and the constitutional moment in which it finds itself.
The functional approach, which requires a focus on the effects concretely produced in society by the category of race, can undoubtedly be used to interpret some of the issues in biomedicine.

3.2. The contribution of race to (in)justice and (in)equality: From blindness to consciousness

As we have seen, the study of genetics has always been enmeshed with racist ideas and all developments in this science should therefore be carefully monitored. A tendency to misappropriate the principles of genetics, as we have seen, continues. In 2018, the American Society of Human Genetics (ASHG) issued a strong statement denouncing attempts to link the science to white supremacy: "Any attempt to use genetics to rank populations demonstrates a fundamental misunderstanding of genetics". It went on to urge scientists to "debunk genetics-based arguments promoting racial supremacy."[52] Even more recently, the society reiterated its strong opposition to efforts that twist genetic knowledge for social or political ends, affirming "the biological reality that we are one people, one species, and one humanity."[53]
More than a decade after leading geneticists argued that race is not a true biological category, the concept continues to be used in many studies. Race, as Troy Duster put it, has been "buried alive."[54]
While the scientific community is trying to steer the debate in the right direction,[55] the law must keep its focus on equality and the compelling public interest of eliminating disparities, shifting the emphasis from race to the very real, and very disturbing, phenomenon of racism, both in its more explicit and more subtle forms. Racism is, first and foremost, not an ideology but a policy, and this is the angle from which it must be opposed.[56]
Racism, for example, has long had a negative impact on the just provision of health care in many countries.[57]

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[52] AMERICAN SOCIETY OF HUMAN GENETICS, *ASHG Denounces Attempts to Link Genetics and Racial Supremacy*, in *American Journal of Human Genetics*, 103(5), 2018, p. 636.
[53] AMERICAN SOCIETY OF HUMAN GENETICS, *Statement Regarding Concepts of "Good Genes" and Human Genetics*, 24 September 2020, at: https://www.ashg.org/tag/statement/.
[54] T. DUSTER, *Buried Alive: the Concept of Race in Science*, in *The chronicle of higher education*, 14 September 2001
[55] Instead of throwing out the categories of race and ethnicity in biomedical research, National Institute of Health (NIH) leaders are calling for the scientific community to develop a consensus on their appropriate use in research: see J. MJOSETH, *NIH leaders call for a consensus on use of race and ethnicity data in biomedical research*, online: https://www.genome.gov/news/news-release/NIH-leaders-call-for-a-consensus-on-the-use-of-race-and-ethnicity-data-in-biomedical-research.
[56] Z. BAUMAN, *Modernity and the Holocaust*, cit., according to whom "Racism is a policy first, ideology second. Like all politics, it needs organization, managers and experts".
[57] See the data collected by the Centers for Disease Control and Prevention in the U.S., "Health of Black or African American non-Hispanic population," available at https://www.cdc.gov/nchs/fastats/black-health.htm. It is necessary to recognize that health inequities are presented differently, according to context specificities: whereas in the EU reports usually focus on inequalities in health between people living in different parts of the Union and inequalities between the most advantaged and disadvantaged sections of the population, in the U.S. health disparities are reported to be associated with race and ethnicity as the primary focus (see E. DOCTEUR, R.A.

For instance, the data on Covid-19, in the United States, show a mortality rate almost three times higher in some groups than in others.[58] These studies show that such situations of inequality do not depend on race as a biological feature, understood as an independent risk factor for disease, but rather as a mediator of structural inequalities resulting from racist policies,[59] a "risk marker of vulnerability, bias or systemic disadvantage".[60]

The policies in question are not, in fact, necessarily explicitly racist, rather many of them create inequality indirectly. Some contemporary theories highlight the very close link between public policies and health equality: a group of scholars at Harvard, for example, has developed the so-called "ecosocial" theory of disease distribution and the construct of "embodiment," referring to how we literally (biologically) incorporate, within our societal and ecologic context, the material and social world in which we live. Relevant factors include food insecurity, poor sanitation and a lack of potable water, discrimination, toxic exposure, but the theory's crucial element is its claim that these exposures are structurally shaped by a society's political economy, political ecology, and social history.[61]

If the above thesis is accepted, the role of law cannot be subordinated to science in the field of health equality: it must move to the fore and has many ways of intervening.

To promote equality, it is not necessary to deny human differences - the latter may, indeed, be important at a biological level -, but it is essential to decouple the concept of race from its purely biological component and to endeavor to understand its full complexity. Race should not enter legal

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BERENSON, *In Pursuit of Health Equity: Comparing U.S. and EU Approaches to Eliminating Disparities*, Robert Wood Johnson Foundation and Urban Institute, available at SSRN: https://ssrn.com/abstract=2462922). Nevertheless, the idea that race in Europe is not irrelevant and should be "viewed as an *absent presence*, something that oscillates between reality and nonreality, which appears on the surface and then hides underground" is addressed by some authors (see A. M'CHAREK, K. SCHRAMM, D. SKINNER, *Technologies of Belonging: The Absent Presence of Race in Europe*, in *Science, Technologies and Human Values*, 39(4), 2014, pp. 459-467).

[58] Data collected in 2020 showed a greater mortality rate among African-American/black individuals compared with Latino and European ancestry/white individuals. See T.L. Edwards et al., Equity in Health: Consideration of Race and Ethnicity in Precision Medicine, in Trends in Genetics, 36(11), 2020, pp. 807-809; R.A. Oppel Jr. et al., The Fullest Look Yet at the Racial Inequity of Coronavirus, in The New York Times, 5 luglio 2020, at https://www.nytimes.com/interactive/2020/07/05/us/coronavirus-latinos-african-americans-cdc-data.html, ant the CDC report at https://www.cdc.gov/coronavirus/2019-ncov/community/health-equity/racial-ethnic-disparities/disparities-illness.html.

[59] J.P. CERDEÑA, M.V. PLAISIME, J. TSAI, *From race-based to race-conscious medicine: how anti-racist uprisings call us to act*, in *The Lancet*, 396, 2020, pp. 1125-1128, meaning that biological differences do not account for the racial and ethnic disparities in Covid-19 illnesses and deaths. The importance of socio-economic and environmental factors is also highlighted by G. OGEDEGBE et al., *Assessment of Racial/Ethnic Disparities in Hospitalization and Mortality in Patients With COVID-19 in New York City*, in *JAMA Netw Open*, 3(12), 2020, e2026881. Other studies show the importance of reporting COVID-19 outcomes by race/ethnicity together with socioeconomic measures including education and occupation, age, and gender (J.T. CHEN et al., *Intersectional inequities in COVID-19 mortality by race/ethnicity and education in the United States, January 1, 2020–January 31, 2021*, Harvard Center for Population and Development Studies, Working Paper Vol. 21, n. 3, 23 February 2021pp. 1-14). Tsai interprets Covid-19 outcomes as a result of racism: *COVID-19 is Not a Story of Race, but a Record of Racism—Our Scholarship Should Reflect That Reality*, in *The American Journal of Bioethics*, 21(2), 2021, pp. 43-47.

[60] J. TSAI, *What Role Should Race Play in Medicine?*, in *Scientific American*, 12 September 2018, at: https://blogs.scientificamerican.com/voices/what-role-should-race-play-in-medicine/.

[61] N. KRIEGER, *Living and Dying at the Crossroads: Racism, Embodiment, and Why Theory Is Essential for a Public Health of Consequence*, in *American Journal of Public Health*, 106(5), 2016, pp. 832-833.

debate as a biological reality, but as a reflection of the cultural and social underpinnings originally used to justify the creation of groups within humanity and their hierarchization, or, in other words, their "othering".[62]

In the field of medicine, existing disparities can be counteracted in at least two ways. The first is by recognizing the real differences between individuals described by science and taking advantage of them to promote forms of equality.

Consider, for example, the field of genetic research, in which some analyses have reported that, despite an acknowledged need to increase the intensity of research in minority groups, most large-scale genetic studies (>70%) have focused on European ancestry populations. Poorly calibrated models can exacerbate disparities because genetic predictors of disease in European ancestry populations do not maintain consistent predictive power in other populations.[63]

It is therefore essential to work out ways of facilitating the recruitment of participants from under-represented populations for clinical studies. More broadly, if the promise of more precise individualized medicine is to be realized, not only more diverse research participants, but also more diverse investigators, funders and editors need to be encouraged, in order to better reflect the demographics of society and its healthcare needs.[64] In this regard, the pluralist spirit of contemporary legal culture can certainly make a valuable contribution to the appreciation and promotion of any differences shown to have a sound scientific basis.

On the other hand, policy makers should consider instruments such as affirmative action to restore balances and equality. Some of the strategies that have been adopted to deal with Covid-19 provide useful topical examples of possible directions.

A few months before the Covid-19 vaccines began to be approved, many countries started to consider which criteria to adopt in choosing who in their populations to vaccinate first. Having established health workers as their absolute priority, more controversial criteria had to be evaluated. In the United States, for instance, the Centers for Disease Control and Prevention and the Advisory Committee on Immunization Practices considered the contentious option of prioritizing the vaccination of Black and Latino people, who have disproportionately fallen victim to Covid-19. The U.S. National Academies of Sciences, Engineering, and Medicine, too, have recommended prioritization of socioeconomically and epidemiologically disadvantaged racial minorities and the World Health Organization has cautioned that "colorblind" allocation frameworks could perpetuate or exacerbate existing injustices.[65]

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[62] Referring to "the process whereby an individual or groups of people attribute negative characteristics to other individuals or groups of people that set them apart as representing that which is opposite to them" (P. ROHLEDER, *Othering*, in T. TEO (ed), *Encyclopedia of Critical Psychology*, New York, 2014).

[63] G. SIRUGO, S.M. WILLIAMS, S.A. TISHKOFF, *The Missing Diversity in Human Genetic Studies*, in *Cell*, 177(1), 2019, pp. 26-31. The same problem can be observed regarding clinical trials in general and has recently been pointed out in relation to anti-Covid vaccines (L.E. FLORES et al., *Assessment of the Inclusion of Racial/Ethnic Minority, Female, and Older Individuals in Vaccine Clinical Trials*, in *JAMA*, 4(2), 2021, e2037640).

[64] G. ADIGBLI, *Race, science and (im)precision medicine*, in *Nature Medicine*, 26, 2020, pp. 1675-1676.

[65] WORLD HEALTH ORGANIZATION, Strategic Advisory Group of Experts (WHO/SAGE), *Values framework for the allocation and prioritization of COVID-19 vaccination*, 14 September 2020, online: https://apps.who.int/iris/bitstream/handle/10665/334299/WHO-2019-nCoV-SAGE_Framework-Allocation_and_prioritization-2020.1-eng.pdf?ua=1 .

As of early March 2021, and despite the efforts shown by the Biden administration,[66] vaccine distribution data demonstrate that the concerns underpinning the above deliberations are all too real.[67]

These positions, however, have been challenged by some medical experts, who are not convinced that prioritization on grounds of race has any sound scientific basis. And some legal experts have questioned whether this "reverse discrimination" could be considered lawful by courts, concluding that it would be more appropriate to adopt vaccine distribution formulas based on factors like geography, socioeconomic status, and housing density that would favor racial minorities de facto, without explicitly referring to race.[68] Another complicating factor is that Black Americans would not necessarily welcome vaccine priority,[69] probably because many do not trust the medical establishment, given the generations of ethical abuse that they have suffered.

A recent South African court decision provides fascinating evidence of the sensitivity of the social issues involved in such matters: the court had to decide on whether to use "race" as one of the criteria in allocating Covid-19 related financial aid, and its ruling, that the "Constitution read as a whole cannot be construed as a libertarian constitution" (as opposed to egalitarian) "or as a race-neutral constitution", was greeted without particular surprise. To ignore race, according to the Court, would be to erase the country's "egregious history in which race overlaid by class and gender was the central determinant of the distribution of resources in our society for more than 300 years of its existence."[70]

The concept of race will not go away simply because it is denied: race should be spoken about in terms of current racism, to combat socioeconomic disparities and to move from a "race blind" to a "race conscious" approach. Race, in its configuration as a multifactorial complex concept, cannot simply be overlooked if real equality is to be built[71]. The political nature of race requires concrete analysis of various domains in order to determine, firstly, when and where using the concept outweighs the costs [72] and, secondly, which political actions can redirect its effects.

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[66] S. GAY STOLBERG, *As Biden Pushes for Racial Equity in Vaccination, Data Is Lagging*, in The New York Times, 9 February 2021, online: https://www.nytimes.com/2021/02/09/us/politics/biden-vaccination-race-data.html.

[67] So far, 41 States in the U.S. have released data on vaccine distribution by race and ethnicity, showing great disparities (https://www.kff.org/coronavirus-covid-19/issue-brief/latest-data-on-covid-19-vaccinations-race-ethnicity/). Of relevance here is that data are essential to prioritizing racial and ethnic equity: as of 1 March 2021, data collected by the CDC showed that race/ethnicity was known for just over half (54%) of people who had received at least one dose of the vaccine (https://covid.cdc.gov/covid-data-tracker/#vaccination-demographic).

[68] H. SCHMIDT, L.O. GOSTIN, M.A. WILLIAMS, *Is It Lawful and Ethical to Prioritize Racial Minorities for COVID-19 Vaccines?*, in *JAMA*, 324(20), 2020, pp. 2023-2024. According to the Authors, public health agencies "should not exacerbate racial divisions" with vaccine allocation decisions, and have an opportunity to become "agents of change toward improving social and racial justice."

[69] In a late November poll by the Pew Research Center, only 42% of Black adults said they would "probably" or "definitely" take the vaccine, compared with 61% of white, 63% of Latino and 82% of Asian adults (https://www.pewresearch.org/science/2020/12/03/intent-to-get-a-covid-19-vaccine-rises-to-60-as-confidence-in-research-and-development-process-increases/).

[70] Case no. 21424/2020, commented by P. DE VOS, *Court ruling on Covid-19 relief criteria affirms that the pandemic is not the 'great equaliser'*, in Daily Maverick, 23 June 2020.

[71] As recently stated: "Dislodgement of race from research may hide still-evident and often egregious episodes of health disparities", see J.P.A. JOANNIDIS et al., *Recalibrating the Use of Race in Medical Research*, in *JAMA*, 325(7), 2021, pp. 623-624.

[72] M.R. JAMES, *The political ontology of race*, cit., pp. 106-134.

4. From yesterday's discrimination to tomorrow's: A.I. as a multiplier or reducer of racial bias?

A.I. technologies are having a significant impact on the debate around the definition of the concept of race, its boundaries of truth, and its role in contemporary societies. These technologies are developing rapidly and their interaction with biotechnologies could very possibly provide immense human health benefits. In this, as in many other fields, "The way we approach AI will define the world we live in".[73] However, as is so often the case with transformative technologies, new opportunities are accompanied by new risks.[74]

One of the many potential pitfalls of artificial intelligence applications - already widely discussed by legal experts - is the existence of biases that inevitably affect the functionality of these new applications of technology. Such biases may concern the data sets used (What characteristics do they include? Where do they come from? How are the data collected and selected?),[75] the algorithms applied (How are they structured? Can they themselves be biased in their operation?), the interaction between datasets and algorithms (Is it possible for bias to emerge, despite the accuracy of the dataset and the correctness of the algorithm?). [76]

This issue is extremely relevant to both fully and partially automated decisions (in the latter, "automation bias" is accompanied by the human tendency to be influenced by the "algorithmically prepared" decision).[77]

Of particular concern to us here is the risk that AI applications either produce new discriminations or reproduce and amplify old ones. The new systems can inherit human racial prejudices and reproduce them in digital technological life, thus reinforcing the role of a category which has no scientific basis. Examples are already familiar: consider the case of self-driving cars that detect pedestrians with different skin tones with different levels of accuracy[78] thereby exposing darker-skinned people to greater risk; the use of criminal risk assessment technology which has led to black individuals

[73] COM (2018) 237 final, Communication from the Commission to the European Parliament, the European Council, the Council, the European Economic and Social Committee and the Committee of the Regions, *Artificial Intelligence for Europe*, 25 April 2018, in www.ec.europa.eu, p. 1.

[74] J. T. O'Brien, C. Nelson, *Assessing the Risks Posed by the Convergence of Artificial Intelligence and Biotechnology*, in Health Security, 18(3), 2020, pp. 219-227.

[75] As highlighted by the EU Commission, "The quality of the data sets used is paramount to the performance of AI systems. When data is gathered, it may reflect socially constructed biases, or contain inaccuracies, errors and mistakes. This needs to be addressed prior to training an AI system with any given data set" (COM(2019) 168 final, Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions, *Building Trust in Human-Centric Artificial Intelligence*, p. 5).

[76] See A. Santosuosso, M. Tomasi, *Diritto, Scienza e Nuove Tecnologie*, 3 ed., Milano, 2021, p. 353.

[77] Cfr. I.J. Skitka, K.L. Mosier, M. Burdick, *Does automation bias decision-making?*, in *International Journal of Human-Computer Studies*, 1999, p. 991.

[78] A. Hern, *The racism of technology - and why driverless cars could be the most dangerous example yet*, in *The Guardian*, 13 March 2019, referring to B. Wilson, J. Hoffman, J. Morgenstern, *Predictive Inequity in Object Detection*, in arXiv:1902.11097, 21 February 2019.

being sentenced to harsher criminal sentences because of a higher risk of recidivism;[79] the companies which have started to use race-detection software to understand how certain customers use their products, who looks at their ads, or what people of different "racial" groups like;[80] the healthcare companies that rely on algorithms that deem Black patients less worthy of critical healthcare than other groups with similar medical conditions.[81]

These examples explain why, as of 2019, in US cities such as San Francisco, Portland, Oakland and Boston, ordinances have been issued prohibiting the use of facial recognition by municipalities and individuals. These decisions came after a MIT study found that facial analysis programs had an error rate of up to 35% for darker skinned women[82] and the wrongful arrest of innocent people had sparked protests in a number of cities.[83] In New York, Amnesty International launched a global campaign to ban the use of facial recognition systems, on the grounds that they exacerbate systemic racism.[84]

The danger is insidious because, in most cases, the technology is not created with discriminatory intent; artificial intelligence systems are assumed to be color blind, since racial information is not usually an input.

Nonetheless, even unintentional or unconscious discrimination can, of course, cause harm. The combination of AI technology and an unjust social system risks both spreading and reinforcing injustice and vesting decisions based on A.I. with a character of neutrality and objectivity that they do not always deserve.[85]

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[79] In 2016 COMPAS the US news organization *ProPublica* reported that COMPAS (Correctional Offender Management Profiling for Alternative Sanctions), an algorithm widely used in the US to guide sentencing by predicting the likelihood of a criminal reoffending, was racially biased. According to the analysis, the system predicts that black defendants pose a higher risk of recidivism than they actually do, and the reverse for white defendants. See J. ANGWIN, J. LARSON, S. MATTU, L. KIRCHNER, *Machine bias: There's software used across the country to predict future criminals. And it's biased against blacks*, in *ProPublica*, 23 May 2016, online: https://www.propublica.org/article/machine-bias-risk-assessments-in-criminal-sentencing. The same risk is highlighted by the EU Commission, according to which: "Certain AI algorithms, when exploited for predicting criminal recidivism, can display gender and racial bias, demonstrating different recidivism prediction probability for women vs men or for nationals vs foreigners" (COM(2020) 65 Final, *White Paper on Artificial Intelligence - A European approach to excellence and trust*, p. 11). Reference goes to S. TOLAN, M. MIRON, E. GOMEZ, C. CASTILLO, *Why Machine Learning May Lead to Unfairness: Evidence from Risk Assessment for Juvenile Justice in Catalonia*,ICAIL '19: Proceedings of the Seventeenth International Conference on Artificial Intelligence and Law, 2019, pp. 83-92.

[80] P. OLSON, *The quiet growth of race-detection software sparks concerns over bias*, in The Wall Street Journal, 14 August 2020, at: https://www.wsj.com/articles/the-quiet-growth-of-race-detection-software-sparks-concerns-over-bias-11597378154.

[81] Z. OBERMEYER, B. POWERS, C. VOGELI, S. MULLAINATHAN, *Dissecting racial bias in an algorithm used to manage the health of populations*, in *Science*, 336, 2019, pp. 447-453.

[82] J. BUOLAMWINI, T. GEBRU, Gender Shades: Intersectional Accuracy Disparities in Commercial Gender Classification, in Proceedings of Machine Learning Research, 81, 2018, pp. 1-15.

[83] A case of faulty facial recognition leading to wrongful arrest is reported by K. HILL, *Wrongfully Accused by an Algorithm*, in *The New York Times*, 24 June 2020, at: https://www.nytimes.com/2020/06/24/technology/facial-recognition-arrest.html.

[84] See the campaign *Ban the Scan in New York* which identifies facial recognition as a threat to "the rights of Black and Brown people" (https://banthescan.amnesty.org/).

[85] J. DANIELS, *'Colorblind' Artificial Intelligence Just Reproduces Racism*, in *Huffpost*, 16 January 2019.

In order to achieve equality, the promotion and protection of difference must be ensured when designing A.I. technologies. Most immediately, perhaps, is the need to address the underrepresentation of women and people of color in technology, and the under-sampling of these groups in the data fed to AI systems.[86]

Only if properly and pluralistically constructed, implemented and interpreted, can AI contribute to a fairer society. According to some studies, the true potential of AI lies in revealing existing biases and thereby motivating societal change and, for example, correcting disparities in health care.[87] If this potential is not met, however, a technology optimized for a small part of the world will circulate and - regardless of the criteria used to identify us - benefit only certain groups of people.

[86] J. BUOLAMWINI, *Artificial Intelligence Has a Problem With Gender and Racial Bias. Here's How to Solve It*, in *Time*, 7 February 2019.

[87] For instance, solutions of algorithmic fairness have been developed to create models designed to produce non-discriminatory predictions by limiting bias with respect to predicted outcomes for protected identities, such as race or gender. See M.D. MCCRADDEN, S. JOSHI, M. MAZWI, J.A. ANDERSON, *Ethical limitations of algorithmic fairness solutions in health care machine learning*, in *The Lancet Digital Health*, 2(5), 2020, E221-E223.

ISSN 2284-4503

Special issue

Incidental findings and the right not to know in clinical setting: Constitutional perspectives

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ABSTRACT: Next generation sequencing (NGS) induces frequent discoveries of incidental findings. This means that, during the sequencing, primary information concerning the alteration in gene for which the sequencing test was ordered goes with other information on different alterations. This problem is first faced by laboratories, followed by clinicians. The core question is whether to inform patients of those alterations. The first part of this paper overviews the guidelines adopted by the scientific community to set incidental findings. References are made to the 2016 European Guidelines for Diagnostic NGS, U.S. Recommendations adopted in 2013 and revised twice, in 2015 and in 2016, Italian Report of Bioethics Committee of 2016, and French Guidelines on secondary findings related to cancer gene of 2018. The second part of this paper discusses the rationale of "the right not to know" and analyses two main frameworks: autonomy and privacy. An attempt is made to consider the issue through different constitutional backgrounds: the U.S. and French notion of autonomy and freedom, and the Italian constitutional background. This paper argues that the right not to know is a negative right comprising a denial, but whose exercise requires positive obligations from clinicians to fulfil an effective and conscious choice. Recalling a famous U.S. debate on negative and positive liberty, a synthesis of the two sides could be managed through a procedural setting of consent, including information about family members. In this sense, the right not to know refers to a constitutional pattern of principles that is not limited to self-determination but entails solidarity and responsibility.

KEYWORDS: Incidental findings; Genome sequencing; Autonomy; Privacy; Negative and positive liberties

SUMMARY: 1. Introduction – 2. Incidental findings in clinical setting: A scientific overview – 3. U.S., Italian and French Guidelines on disclosing secondary findings: Scope of application and working procedures – 4. *(continue)* and contents: Types of variations and actionability, the clinician's viewpoint – 5. *(continue)* the patient's viewpoint: The uncertainty of information – 6. The right not to know: Legal basis and conceptual objections. –7. Theoretical frameworks between autonomy and privacy – 8. Constitutional backgrounds between negative and positive liberty – 9. Consent and the right of self-determination in Italian constitutional

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framework – 10. *(continue)* The right not to know under Italian constitutional law – 11. The positive side of the right not to know: Consent – 12. Conclusions: Double consent as a nudge.

1. Introduction

"The right not to know" questions some deeply rooted principles underlying constitutionalism – the idea to change our lives through human will and action. The evolution of technology and science faces borders, where knowledge refusal, at the given state of understanding and depending on intimate and private beliefs, probably is the best chance to organise a worthy life. Concurrently, such an intimate decision is inextricably intertwined with others, family members whose relation could be good or bad, as always in life. This paper argues that exercising "the right not to know" reflects autonomy but refuses isolation and calls for solidarity and responsibility. In this framework, there is a positive obligation of public authorities to set appropriate measures through which everyone can manage his faith and expectations for how his life is supposed to be.

This paper explores incidental findings in a clinical setting. Managing them raises different problems in both clinics and research, and even if the borders between research and clinic are increasingly blurring, we concentrate only on clinical aspects.[1] Also, this paper does not discuss issues concerning minors and third parties other than family members.

[1] The main differences between incidental findings in clinical and research settings are as follows: genome sequencing for clinical diagnosis primarily entails protecting the individual patient's health, so an examination ordered for clinical reasons involves evaluations on prevention or treatment; the interests of others, particularly family members, are much more compelling in clinical practise than in research activity; genomic analysis performed for health research sometimes lacks the same sensitivity and quality criteria (depth and coverage) as that performed in a healthcare context. The main point for incidental findings in research seems to be the opportunity or duty of recontact, depending on the scale of the study, the object - rare diseases or not - and the formula of initial consent. See J. VIBERG, M.G. HANSSON, S. LANGENSKIÖLD, P. SEGERDAHL, *Incidental Findings: The Time Is not yet Ripe for a Policy for Biobanks*, in D. MASCALZONI (ed.), *Ethics, Law and Governance of Biobanking*, Heidelberg, 2015, 121-131, and, in Italian literature, M. TOMASI, *Genetica e Costituzione*. *Esercizi di eguaglianza, solidarietà e responsabilità*, Napoli, 2019, 245 ff. On the right not to know in research, S. M. WOLF, J. PARADISE, C. CAGA-ANAN, *The Law of Incidental Findings in Human Subjects Research: Establishing Researchers' Duties*, in *Journal of Law, Medicine and Ethics*, 36, 2, 2008, 361-383; B.M. KNOPPERS, *From the Right to Know to the Right Not to Know*, in *Journal of Law, Medicine and Ethics*, special issue no. 1, 2014, 1-10, who notes that "no return" policies adopted by large population genome studies, for example, P3G, on 23 national biobanks, have been integrated with new options of consent upon recontact of participants, because the increasing use of WGS caused the discovery of unsolicited information such as rare mutations or clinically significant incidental findings, blurring the distinction between research and clinical contexts. Nevertheless, the borders between research and clinics in the matter still exist. See *European Society of Human Genetics' Guidelines for diagnostic next-generation sequencing*, in *European Journal of Human Genetics*, 24, 2016, 2-5, 5, on "Distinction between Research and Diagnostic", Statements no. 32-38. Recently, the American College of Medical Genetics and Genomics gave a statement to forbid using their clinical recommendations for incidental findings for general population screening because any of the ACMG SF v2.0 genes, as quoted later, have uncertain penetrance when identified in asymptomatic individuals (e.g. SCN5A and Brugada syndrome). In the absence of penetrance data that can only be obtained through robust genotype-phenotype correlation, the medical ethical principle of nonmaleficence should dominate, in *The use of ACMG secondary findings recommendations for general population screening: a policy statement of the American College of Medical*

The first part of this paper overviews current guidelines and recommendations adopted by the scientific community to set incidental findings. References are made to the 2016 European Guidelines for Diagnostic Next Generation Sequencing (NGS), U.S. *Recommendations* adopted in 2013 and revised twice, in 2015 and in 2016, Italian Report of Bioethics Committee of 2016, and French *Guidelines* on secondary finding related to cancer gene of 2018. A comparison is made on the procedures adopted to define the guidelines and to their content by considering clinicians' viewpoints (types of variations and actionability) and patients' viewpoints (right not to know). In this first part we will use both the acronyms IFs, incidental findings, present in the Italian Report, and SFs, secondary findings, where U.S. *Guidelines* since the 2015 version and French *Guidelines* refer specifically to secondary findings, as we will better explain later.

The second part of this paper discusses the rationale of "the right not to know" by analysing two main frameworks having a great echo between scholars: autonomy and privacy. Subsequently, an attempt is made to consider the issue through different constitutional backgrounds: the U.S. and French notion of autonomy and freedom, and the Italian constitutional framework. Constitutional traditions are relevant because the clinician–patient relationship mirrors an individual's position in society and the relation with power. The common tread chosen is the negative and positive way "the right not to know" is framed. The two aspects, in our opinion, do not create dichotomy, but coexistence – the positive side setting the condition to fulfil the negative one. Hence, this paper argues that "the right not to know" is a negative right comprising a denial, but whose exercise requires positive obligations from clinicians to fulfil an effective and conscious choice. Recalling a famous U.S. debate on negative and positive liberty, synthesising the two sides could be managed through a procedural setting of consent, including information about family members. In this sense, "the right not to know" refers to a constitutional pattern of principles that is not limited to self-determination but entails solidarity and responsibility.

2. Incidental findings in clinical setting: A scientific overview

Next Generation Sequencing[2] and the increasing use of multi-panel analysis in clinical practise to discover genes responsible for main diseases induces frequent discoveries of incidental findings. This means that, during the sequencing, primary information concerning the alteration in gene for which the sequencing test was ordered goes with other information on different alterations. Geneticists

Genetics and Genomics (ACMG), in *Genetics in Medicine*, 2019 (21), 1467-1468, and the ongoing debate that followed. In contrast, French *Guidelines*, as quoted later, 1736, 1738, concern both somatic analyses performed in healthcare context for research, and the provided consent forms too. For a recent example of management in research, see le dossier *Comment donner suite aux découvertes fortuites significatives. Lignes directrices sur l'application de l'article 3.4 EPTC* 2 (2018), *Group consultative en éthique de la recherche*, 2019, in https://ethics.gc.ca/fra/incidental_findings.html.

[2] See the introduction of European Society of Human Genetics' Guidelines for diagnostic next-generation sequencing: "Next generation sequencing (NGS) allows for the fast generation of thousands to millions of base pairs of DNA sequence of an individual patient. The relatively fast emergence and the great success of these technologies in research herald a new era in genetic diagnostics. However, the new technologies bring challenges, both at the technical and in terms of data management, as well as for the interpretation of the results and for counselling", cit., 2.

usually divide IFs into different categories: fortuitous or secondary. Fortuitous findings are completely unexpected, accidental and non-predictable results, while secondary findings are predictable results, i.e. they can be expected as a part of the results, even if they are unrelated to the primary research of alteration.[3] The problem is first faced by laboratories, followed by clinicians. The core question is whether to inform patients of those alterations.

IFs are well known in the fields of medicine other than genetics. For example, radiographs for a particular anatomical focus can report abnormal findings in regions not indicated as the primary reason for investigation. Although it is generally argued that there is a right of the patient to know this information and a duty of the clinician to report them to protect the patient's health, the same pattern may not be suitable for genome IFs. Many differences exist between radiographic images and genetic information regarding predictability of the information and interpretation, both technically and clinically.[4] Furthermore, patient could avoid obtaining genetic information for many reasons. Learning to be at risk of genetic disease may induce anxiety, depression, changes in self-image, reduced capacity to organise the future or even stigmatisation and discrimination.[5] Moreover, the desire for ignorance could rely on economic and social conditions, concerns about the costs of therapies, or the fear of being a burden to others.

Few recommendations have been adopted in Europe to manage IFs, and scientific publications have shown variabilities in European practices for transmitting incidental information both from laboratories to clinicians and from clinicians to the patient. A need for common guidelines emerged to help molecular laboratories and clinician physicians harmonise their practice, particularly for cancer-related genes, and to assure equal access and uniform service across Europe.[6]

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[3] We will see later that U.S. *Guidelines* and French *Guidelines* refer to secondary findings, while the Italian Bioethics Committee use the term "incidental findings" because it is common in international debate: Report *Gestione degli "incidental findings" nelle indagini genomiche con le nuove piattaforme tecnologiche*, 17 marzo 2016, http://bioetica.governo.it/media/1803/p123_2016_incidental_findings_it.pdf, 11, note no. 23.

[4] See French *Guidelines for reporting secondary findings of genome sequencing in cancer gene: the SFMPP recommendations*, in *European Journal of Human Genetics,* 26, 2018, 1732-1742, spec. 1737, admitting that the sensitivity of variant detection may vary among genes according to the coverage and depth of the sequencing process. Thus, the coverage and depth at the position of the gene of interest *will differ from that of genes secondarily and deliberately studied*. This concern could induce misinterpretation of the results by the clinician and/or the patient, such as the absence of a pathogenic variant in secondarily studied genes. Therefore, the French *Guidelines* suggest reporting the conditions of sequencing and analysis of secondary data if it differs from that of the primary data. Similarly, see European Society of Human Genetics' Guidelines, cit., "The implications of diagnostic test based on NGS depend on the procedures, platforms, filtering processes and data storage used in the laboratory. It is crucial that the referring physician is fully informed about the limitations and possible unfortunate effects of a genetic testing".

[5] R. CHADWICK, *The Philosophy of the Right to Know and the Right Not to Know*, in R. CHADWICK, M. LEVITT, D. SHICKLE (eds.), *The Right to Know and the Right Not to Know. Genetic Privacy and Responsibility*, Cambridge, 1997, 13-22, spec. 18.

[6] French *Guidelines*, cit., 1733, quoting D. F. VEARS, K. SENECAL, P. BORRY, *Reporting practices for unsolicited and secondary findings from next-generation sequencing technologies: Perspectives of laboratory personnel*, in *Human Mutation*, 38, 2017, 905-911, for variability in transmitting information from laboratories to clinicians, and J. Y. HEHIR-KWA, M. CLAUSTRES, R. J. HASTINGS, C. VAN RAVENSWAAIJ-ARTS, G. CHRISTENHUSZ, M. GENUARDI et al., *Towards a European consensus for reporting incidental findings during clinical NGS testing*, in *European Journal of Human Genetics*, 23, 2015, 1601-1606, from clinicians to patients. For the need of common guidelines, see C. G. VAN EL, M. C. CORNEL, P. BORRY, R. J. HASTINGS, F. FELLMANN, S. V. HODGSON et al., *Whole-genome sequencing in*

ISSN 2284-4503

Here the issue is addressed by analysing different sets of recommendations adopted in the U.S., Italy, France and at the European level by the European Society of Human Genetics. It will be useful to start from the European level, which demonstrates a lack of common vision for IFs. The 2016 European Society's Guidelines on Diagnostic NGS recognise that the main implication of a diagnostic test based on NGS is the chance of unsolicited and secondary findings. To face the problem, under the paragraph "Informed consent and information to the patient and clinician", Statement no. 9 suggested focusing on the gene panel under investigation because the chance of unsolicited findings in a gene panel is very low and mainly depends on the genes involved. However, if the unsolicited information occurs, *choice is given to each laboratory, institute or to authorities at the national level*, *whether patients are offered opt-in, opt-out options* to get additional information. The only recommendation is that laboratories disclose the chance of unsolicited findings and set up "unsolicited and secondary finding protocols", which must agree with the decision of an ethical committee. Each protocol should specify whether unsolicited findings are reported and, in that case, provide for pre-test genetic counselling, including a discussion on both expected results and the potential for unsolicited and secondary findings, local policies assuring clear dissemination for patients.[7] Overall, adopting opt-in and opt-out policies on IFs is discretionary. A definitive settlement of the interests at stake has not been established, and the balance between the task to report and the desire of the patient remains open. We will see that this setting has been discussed at the national level on both sides of the Atlantic.

3. U.S., Italian and French Guidelines on disclosing secondary findings: Scope of application and working procedures

After a strong scientific debate in 2013, the American College of Medical Genetics and Genomics adopted guidelines (in the following, U.S. *Guidelines*) concerning secondary findings.[8] For their organic and influential attempt to set the issue, U.S. *Guidelines* induced discussion in the scientific

healthcare: Recommendations of the European Society of Human Genetics, in *European Journal of Human Genetics*, 21, 2013, 580-584, and M. CLAUSTRES, V. KOZICH, E. DEQUEKER, B. FOWLER, J.Y. HEHIR-KWA, K. MILLER et al., *Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and molecular genetic)*, in *European Journal of Human Genetics*, 22, 2014, 160-170. Many governments have drawn up plans to develop and generalise the use of large-scale genetic analysis, such as the UK (100,000 Genomes Project), Iceland, Estonia (Estonian Genome Project), France (Genomic Medicine France 2025), The Netherlands (Genome of The Netherlands Project), and Germany (National Genome Research Network). In this dynamic, it is likely that IFs become frequent and a harmonisation of practices to better care for the patient has become necessary.

[7] *European Society of Human Genetics' Guidelines*, cit., 3-4, Statements no. 10, 11 and 12, under "Informed consent and information to the patient and clinician"; under "Reporting": all pathogenic (class 5) and likely pathogenic (class 3) variants have to be reported. Whether or not Unclassified Variants (UVs. class 3) are reported will depend on local practice, which has to be clear for the laboratory scientist and the referring clinician. Statement no. 29 re-clarifies that laboratories should have a clearly defined protocol for addressing unsolicited and secondary findings before launching test.

[8] *ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing*, published on 20 June 2013, in *Genetics in Medicine*, 15, 7, 2013, 565-574, so called ACMG SF v1.0 or "ACMG 56".

community. They have been revised in 2015[9] and 2016[10] and, in the last version, they listed 59 medically actionable genes recommended for return in clinical genomic sequencing (four genes were added to the first list and one removed).

In Europe, no common approach was observed. In Italy, since 2007 the Italian Data Protection Authority gave general provisions on genetic data processing for clinical and research purposes. The aim of this regulation, as defined by the Italian Data Protection Code, was to govern all stages of genetic data processing, from the initial information, to consent, communication and dissemination. The problem of unexpected results was addressed within the more general definition of data processing.[11] In 2016, the National Committee for Bioethics followed the American debate, adopting a specific *Report on Management of Incidental Findings in genomic sequencings with new technological platforms*. As U.S. *Guidelines*, to define IFs, the Committee referred to the 2013 report of the Presidential Commission for the Study of Bioethical Issues, which divided the findings into primary (related to the primary investigation), secondary (anticipable results, so results that the sequencing is looking for) and discovery (non-anticipable results, fortuitous).[12]

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[9] *ACMG policy statement: Updated recommendations regarding analysis and reporting of secondary findings in clinical genome-scale sequencing*, in *Genetics in Medicine*, 17, 1, 2015, 68-69.

[10] *Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): A policy statement of the American College of Medical Genetics and Genomics*, published on 17 November 2016, in *Genetics in Medicine*, 19, 2, 2017, 249-255, so called ACMG SF v2.0 or "ACMG 59". Original version of 2013 uses the term "incidental findings". In 2015 the *Recommendations* were first revised, adopting the term "secondary findings" "because these genes are intentionally being analysed, as opposed to genetic variations found incidentally or accidentally"; the shift in terminology, it is said, also maintained consistency with a recommendation by Presidential Commission on Bioethical Issues, C. WEINER, *Anticipate and communicate: Ethical management of incidental and secondary findings in the clinical, research, and direct-to-consumer contexts*, December 2013*, Report of the Presidential Commission for the Study of Bioethical Issues*, in *American Journal of Epidemiology*, 180, 2014, 562-564.

[11] The Italian Data Protection Authority ("Garante per la Privacy", DPA in the following) is an independent administrative authority established by law no. 675 of 31 December 1996 and regulated subsequently by the Personal Data Protection Code, Legislative Decree no. 196 of 30 June 2003. Before the entry into force of the EU Data Protection Regulation no. 2016/679, the processing of genetic data under Italian law was governed by a so called "general authorisation" issued by the DPA, pursuant to Art. 90 of Legislative Decree no. 196 of 2003, under which the requirements and conditions laid down in the authorisation were the only cases in which processing was allowed. The position and force of the authorisation in the hierarchy of sources gave rise to a wide-ranging debate concerning its assimilation to the law. The first version of the general authorisation dates back to 2007 and was significantly amended in 2011, with renewals up to 2016 (www.garanteprivacy.it/web/guest/home/docweb/-/docweb-display/docweb/5803688). As will be seen in more detail below, one of the most significant changes introduced since 2011 concerned precisely the possibility of allowing the processing of genetic data carried out to protect the health of family members without the consent of the data subject, and the disclosure to family members of genetic data indispensable to prevent harm to their health. Legislative Decree no. 101 of 10 August 2018 amended the Code and established that the Italian DPA is the supervisory authority responsible for monitoring application of the EU General Data Protection Regulation, pursuant to its Art. 51. The general authorisation no. 8 of 2016 has been replaced by the no. 146 of 2019, www.garanteprivacy.it/home/docweb/-/docweb-display/docweb/9124510 (last visited on 19 April 2021).

[12] Report *Gestione degli "incidental findings" nelle indagini genomiche con le nuove piattaforme tecnologiche*, 17 marzo 2016, cit.; the Italian National Committee of Bioethics refers to statements of its previous report of 2010, adopted together with the National Committee for Biosecurity, Biotechnologies and Life Sciences, *Test*

An attempt to generalise secondary findings in genome sequencing has been made in France. In 2017, the French Society for Predictive and Personalised Medicine (*Société Française de Médecine Prédictive et Personnalisée*, SFMPP in the following) organised *ad hoc* working groups to identify secondary findings (*données secondaires*) implications and treatments, and in 2018, the French *Guidelines* were published (in the following also SFMPP *Recommendations*).[13] They were expressed regarding an extensive European genome project and aimed at providing a first step towards standardised guidelines in France and all Europe.

We will focus briefly on the scope of application and the procedure of adoption of the U.S., French *Guidelines* and the Italian Bioethics Committee's Report.

Concerning the scope of application, all documents aim at giving indications, without limiting or substituting the judgment of the clinician, whose adherence to recommendations is voluntary. The U.S. *Guidelines* self-define as an "educational resource", and both U.S. and French text refer to the professional, clinical and ethical evaluation of the practitioner, depending on the specific clinical circumstances presented by the individual patient.[14]

Regarding the adoption procedure, the Italian Report, due to the composition and tasks of the Bioethics Committee, does not enter detailed discussion on the type of genetic variations, while the U.S. and French *Guidelines* are the results of a participatory procedure, between the medical community in the U.S., and more extensively between clinicians, experts in bioethics, philosophers, sociologists and lawyers in France, both aiming at classifying variations and their management. In detail, the U.S. list of pathogenic variations was composed following a bottom-up process. The Secondary Findings Working Group (SFWG) created by ACMG and other colleagues collected information in a nomination form. Completed forms were reviewed in a study section-like model and by another working group. Each form was presented in an SFWG meeting or conference called by the submitter or a designee and discussed. After discussion, SFWG members voted on whether to accept the submitter's recommendation. Nominations recommended by the SFWG were sent to ACMG Board of Directors with a summary of the SFWG discussion, voting outcome, and a recommendation for the suggested change to the SF minimum list.

In France, from June 2016 to May 2017, the SFMPP established a working group of 47 experts to elaborate guidelines for managing SFs for cancer-related genes. The experts were divided into two subgroups regarding ethical and legal questions and medical expertise. The ethical and legal group comprised 17 members, psychologists, sociologists, ethicists, methodologists, lawyers and members of patients' associations, to elaborate general recommendations on information related to patients

genetici di suscettibilità e medicina personalizzata, in http://bioetica.governo.it/media/4045/test-genetici-sulla-suscettibilita.pdf, confirming the distinction between research and clinical activity, and the distinction between minors and adults.

[13] See *Guidelines for reporting secondary findings of genome sequencing in cancer gene: The SFMPP recommendations*, in *European Journal of Human Genetics,* 26, 2018, 1732-1742.

[14] *ACMG SF v2.0*, cit., 249; the *SFMPP Recommendations*, cit., 1739: "The present recommendations are not meant to substitute for the clinical and ethical judgement of clinician, but they are proposed as a basis for reflection to help with the prescription and the results of the genomic analyses in a homogeneous way. These recommendations are not a substitute for a personal bibliographic watch that is an integral part of the clinician's work in a context of continuous improvement of medical knowledge due to the variety of cancer-predisposition syndromes and the technical complexity […]".

and consent, and to provide informed consent forms and information media tools. The medical expertise group comprised 30 members, oncologists, clinical geneticists, molecular biologists, and cancer gene experts, who provided independent evaluation and classification of cancer genes for cancer risk and actionability.[15] To homogenise the results, evaluation criteria were designed on the models of the ACMG, with the aim of creating a common global standard. Both U.S. and French *Guidelines* are presented as provisional text, calling for updates and research on the list of variations and psychological impact of returning SFs.

4. *(continue)* and contents: Types of variations and actionability, the clinician's viewpoint

As we said, due to the composition and tasks of the National Bioethics Committee, the Italian Report did not aim at distinguishing types of variations, while both U.S. and French *Recommendations* included a detailed list of variants. Both considered differences in genetic variations: pathogenic (causative) mutations, polymorphisms, variations that can be found also in healthy people. Both *recommended restricting the report of SFs to pathogenic variations*, providing a list considered "minimal", and open to regular update.[16]

Moreover, both rely on the concept of *actionability* (*données secondaires actionnables* in French) or clinical utility. We will see that actionability is the comet guiding clinicians' perspective.

Broadly speaking, utility measures the personal benefit that someone has from an intervention, outcome, product or process, and medical clinical utility describes the relevance and usefulness of an intervention in patient care.[17] The meaning of "clinical utility" adopted by both U.S. and French *Guidelines* refers to the existence of preventive measures or treatments.[18] Since 2013, the U.S.

[15] In detail, the ethical and legal group comprised three psychologists, one sociologist, four ethicists, two methodologists, four lawyers and three representatives of patient associations (BRCA France, the Vaincre les Maladies Lysosomales association and the Association pour la Prévention, Traitement Etude des Polyposes Familiales); the medical group eight oncologists, eleven clinical geneticists, three molecular biologists and eight cancer gene experts.

[16] See the *SFMPP Recommendations*, cit., 1739. *ACMG SF v2.0*, cit., 251, adopting the classification of variants in S. RICHARDS, N. AZIZ, S. BALE ET AL., *ACMG Laboratory Quality Assurance Committee. Standards and guidelines for the interpretation of sequence variants: A joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology*, in *Genetics in Medicine*, 17, 2015, 405-424.

[17] L. J. LESKO, I. ZINEH, S.-M. HUANG, *What Is Clinical Utility and Why Should We Care?*, in *Clinical Pharmacology & Therapeutics*, 88, 6, 2010, 729-733.

[18] It seems useful to quote *ACMG SF v2.0* on actionability: *"*Initially, medical actionability was evaluated according to a semiquantitative metric that included the following major adjudication criteria: severity of disease/nature of the health threat; likelihood of the disease/health threat materializing (i.e., penetrance); efficacy of specific intervention(s); and overall strength of the current knowledge base about the gene/condition. In July 2015, a fifth criterion was added: acceptability of the proposed intervention based on its risks and benefits. The SFWG acknowledged the inherent subjectivity and difficulty of rating any given intervention as it applies to an individual but voted unanimously in favour of adding this fifth criterion". An example on evaluation of a pathogenic variation could be read in the same *Guidelines*, concerning genes added to the original list: ATP7B is associated with autosomal-recessive Wilson disease (MIM 277900). Morbidity among homozygotes directly correlates with copper deposition in the liver, brain, and eye. The disease is progressive, and, if left untreated, premature death is likely. In some cases, liver failure may be the presenting

Recommendations admitted that there was much to be learned about disease predictability from genomic testing, particularly in asymptomatic individuals. A debate occurred between some arguing that IFs should not be reported at all in clinical sequencing until there is strong evidence of benefit, and others advocating that variations in any all disease-associated genes could be medically useful and should be reported. The working group admitted that there was insufficient evidence about benefits, risks and costs of disclosure to make evidence-based recommendations, a consensus on the listed variations having been found on clinical experience largely from patients with symptoms or positive family histories. The conclusion was that: "Given the low prior probability that an individual has a monogenic disorder that could be identified incidentally through exome or genome sequencing, we recommended that only variants with a higher likelihood of causing disease should be reported as IFs although we recognise that there are limited data available in many cases to make this assessment". So, the minimal list was limited to variants "where there is strong evidence of high probability of severe adverse medical outcomes that can be prevented or ameliorated by existing modalities".[19] In this sense, actionability is the leading principle driving IFs information, and it mirrors the bioethics principle of benefit, *bienfaisance*, in French.[20]

French *Guidelines* also define actionability as availability of screening or prevention strategies, risk evaluation (severity, penetrance and age of disease onset), and level of evidence from published data. Under these criteria, genes have been divided into three classes and 60 genes were listed in class 1, as actionable pathogenic variants.[21] There is an important overlap between the ACMG and SFMPP lists of actionable genes concerning cancer, except for one or two additional genes classified as class 1.

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sign. Given its long recognition as a Mendelian disorder, it is reasonable to consider Wilson disease to be at least relatively highly penetrant. Expressivity is variable. Treatment for Wilson disease involves administration copper chelating agents and/or zinc to block intestinal absorption of copper; treatment is extremely effective when administered prior to the onset of symptoms. Sanger sequencing of the ATP7B gene is considered confirmatory in asymptomatic patients. In symptomatic patients, in addition to Sanger sequencing, the results of serum ceruloplasmin, serum copper concentration and 14-hour urine copper excretion can be diagnostic. The ClinGen Actionability scoring process generated a high actionability score of 10/12 for copper chelation and zinc therapy in the treatment of ATP7B-associated liver disease and/or neuropsychiatric disease. Based on this evidence, the SFWG voted unanimously to add ATP7B to the SF list for the recessive state in which two KP/EP variants are detected in trans through GS. The benefit principle is also based on the recent ACGM, *Patient re-contact after revision of genomic test results: Points to consider—A statement of the American College of Medical Genetics and Genomics*, in *Genetics in Medicine*, 21, 4, 2019, 769-771.

[19] AMGC, *Incidental findings in clinical genomics: A clarification* in *Genetics in Medicine*,15, 8, 2013, 664-666, developing considerations of 2013 *AMGC SF v1.0*, cit.

[20] Three conditions should exist to have benefit: discovery must be scientifically validated; it could be meaningful for the patient health; and there exist a therapy or a preventive behaviour. For general references, see M. TOMASI, *Genetica e Costituzione*, cit., 33-35.

[21] In detail, for class 1 genes (n = 36), delivering the information on SFs was *recommended*; for class 2 genes (n = 5), delivering the information remained *questionable* because genes include significant risk and detection/prevention possibilities, but the literature data or level of evidence seems too low to measure the real benefit of an intervention in an asymptomatic context; and for class 3 genes (n = 19), delivering the information on SFs was *unrecommended* because of the moderate risk of cancer and/or prevention or limited or non-existent therapeutic possibilities.

Overall, for clarity, in both U.S. and French *Guidelines*, disclosing secondary findings is now recommended, but not binding, for actionable pathogenic variants, taking advantage of the patient's health. In contrast, a pathogenic variation of an incurable disease will be uncommunicated. The same is true for variations of uncertain or unknown significance, whose evidence should not be given to the practitioner (first step) or the patient (second step), because it can involve misinterpretation in the results, being unnecessary for the patient's healthcare.[22]

In Italy, the Data Protection Authority general authorisation no. 8 of 2016 adopted the narrow concept of clinical utility, defining unexpected findings in the context of genetic testing *consent* and *communication* as those that "represent a concrete and direct benefit in terms of therapy or prevention or awareness of reproductive choices".[23] But the Italian Committee of Bioethics Report, as a soft law source specifically focusing to incidental findings, adopted a different way. First, as we anticipated, the Report did not refer to an explicit list of variations, as cancer-associated genes. Second, the Committee adopted a smoother and descriptive approach, illustrating different meanings of clinical utility, the traditional stricter view, present in the U.S. *Guidelines* and in the following French ones, where there are preventive measures or treatment, and a broader sense, including wider considerations on situations of non-immediate interventions, or non-existing treatment or non-pathogenic information. This broader meaning of clinical utility implies a corresponding wider assessment of benefits and risks, involving the patient, family or third-person interests, and concerning all decisions where a choice can occur, like reproductive choices, assurance schemes, or other planning of life. From this perspective, all information concerning an opportunity of choice may be reported. The Committee observed that there is a general agreement in the literature on disclosure of actionable findings, while communication of uncertain results or results with no prevention or treatment available is discussed. In the end, the Committee *suggests leaving to the patient, during pre-test and post-test counselling, the choice whether to have information and the kind of information*, i.e. only pathogenic variations or even variations related to untreatable disease. In contrast, the Committee agrees that unknown variations or variations with uncertain significance should not be reported.[24] By this way, an extensive discretionary is given to the patient, while a relatively less intense duty of qualification of clinical utility remains to the doctor.

[22] See also AMCG, *Incidental findings in clinical genomics: A clarification*, cit.: "We agree that variants of unknown significance, variants associated with low or unknown penetrance, and variants associated with disorders not currently amenable to intervention should not be reported". For incidental findings of unknown significance, the SFMMP works assessed that additional follow-up tests or procedures could be risky and costly.

[23] Italian Data Protection general authorisation no. 8 of 2016, cit., par. 6, and now in the same way no. 146 of 2019, par. 4.5. sub 3.

[24] Report *Gestione degli "incidental findings"*, cit., 18: "si rispetti, nella acquisizione del consenso informato, il diritto all'autodeterminazione del paziente e pertanto sia lasciata al consultando, una volta che ha compreso la differenza tra le diverse tipologie degli IF, la scelta di decidere quali risultati conoscere. Il paziente deve essere libero di scegliere se rifiutare le informazioni sugli IF, oppure ricevere solo informazioni relative alle patologie prevenibili o trattabili, o ancora conoscere anche i dati che riguardano condizioni patologiche al momento non prevenibili né curabili", and note no. 39 on uncertain variations: "Non è opportuno invece comunicare le varianti di significato incerto, né le suscettibilità per i motivi già ricordati". In sum, there is a difference between the strict clinical utility for consent and communication of the DPA general authorisation and the broad clinical utility of the Bioethics Committee's Report, where the first is a mandatory source and the second is not. This is a symptom of the lack of clarity and sensitivity on this issue. For the sake of completeness, the

Clearly, statements on actionability or clinical utility, and their consequences on information, put the attention on clinicians' duties and tasks. Now, it is necessary to investigate the guidelines from the patient's viewpoint, and the U.S. debate has been significant again.

5. *(continue)* the patient's viewpoint: The uncertainty of information

From the patient's viewpoint, the U.S. original version of *Guidelines* in 2013 recommended a *mandatory disclosure of actionable secondary findings*, but this statement was revised in 2015, in a relatively short time. Starting from the beginning, in 2013 the working group recognised that denying a preference whether to receive information may be seen to violate existing ethical norms regarding the patient's autonomy and the "right not to know" genetic risk information. However, they felt that "clinicians and laboratory personnel have *a fiduciary duty to prevent harm* by warning patients and their families about certain IFs and that this principle supersedes concerns about autonomy, just as it does in the reporting of IFs elsewhere in medical practice".[25] In detail, the debate on reporting IFs was presented as follows: While genetic libertarians believe that patients have the right to full and complete accounting of all possible risks conveyed by both established and novel variants, or even variants of unknown significance in disease genes, genetic empiricists believe that there is insufficient evidence about the penetrance of most pathogenic variants in the general population to warrant the sharing of any IFs, and that it is irresponsible to create the psychological burdens of being a "patient in waiting" or to expose patients to iatrogenic harm of possibly unnecessary surveillance or diagnostic testing.[26] The fair balance comprised precisely reporting only the listed variations, without seeking preference from the patient and family. So, the duty to protect health prevails when results convey the near certainty of an adverse yet potentially preventable medical outcome. Not returning under these conditions was considered unethical.[27]

DPA general authorisation no. 8 of 2016 and the ongoing no. 146 of 2019 both refer to an extensive meaning of utility in another stage of data processing, during the pre and post-test genetic counselling. The counsellor is called upon to help the person concerned to independently take the most appropriate decisions, taking into account genetic risk, family aspirations and ethical and religious principles, to achieve the best possible adaptation to the disease (see par. 4.4. of the ongoing authorisation).

[25] *ACMG SF v1.0*, continuing: "whenever clinical sequencing is ordered, the ordering clinician should discuss with the patient the possibility of incidental findings, and that laboratories seek and report findings from the list described in the [list] without reference to patient preferences. Patients have the right to decline clinical sequencing if they judge the risks of possible discovery of incidental findings to outweigh the benefits of testing".

[26] *ACMG SF v1.0*, quoting J. M. KWON, R. D. STEINER, *I'm fine; I'm just waiting for my disease: The new and growing class of presymptomatic patients*, in *Neurology*, 77, 6, 2011, 522-523.

[27] AMCG, *Incidental findings in clinical genomics: A clarification*, cit.: "The rationale for our recommendations was that failure to report a laboratory test result conveying the near certainty of an adverse yet potentially preventable medical outcome would be unethical. A common objection to this recommendation has been that some genetic test results *might not be predictive* of disease, either because of incomplete penetrance or because of the occurrence of variants of unknown significance. The recommendations, however, explicitly focus only on unequivocally pathogenic mutations in genes in which pathogenic variants lead to disease with very high probability and cases in which evidence strongly supports the benefits of early intervention. We

Three points should be clarified. First, the 2013 U.S. *Guidelines* version clearly explained that concern and uncertainty surrounding IFs depend on a lack of empiric data, both on predictability and patients' behaviours. Second, this position plainly rejected genetic "exceptionalism", recalling that reporting IFs is a form of screening that had a long history in clinical medicine, where disease to one system could call for other systems' testing in multisystem diseases.[28] Third, it relied strongly on the clinical-patient relationship. The patient has already been presented to the medical care system and has been evaluated by a clinician who is familiar with the patient's conditions and family history. The clinician-patient interaction is the appropriate place for extensively evaluating the opportunity of a specific genetic testing, and for explaining and discussing IFs eventuality.[29] Patients' opt-out option is not completely denied in this pattern, but is conceived at the beginning of the procedure. The patient, conscious of the possibility of extra findings, declines clinical sequencing if the risks of possible incidental discoveries outweigh the benefits of testing.

Refusing opt-in opt-out induced a controversial debate.[30] In 2015, the *Guidelines* were first revised, introducing the possibility to decline SFs through "the right not to know". The version of 2016

agree that variants of unknown significance, variants associated with low or unknown penetrance, and variants associated with disorders not currently amenable to intervention should not be reported".

[28] *ACMG SF v1.0*, "When patients complain of symptoms in the digestive system, the well-trained physician examines cardiac and respiratory systems as well, both for clues to a multisystem disease and to incidentally discover any unrelated signs. When radiographs are read for a particular anatomical focus, the radiologist scans the entire radiograph and also reports on abnormal findings in regions not indicated as the primary reason for the study. In these situations, the patient has already presented to the medical care system, has been evaluated and is under the care of a clinician […]".

[29] See also AMCG, *Incidental findings in clinical genomics: A clarification*, cit., where the clinician, and his or her consultant, i.e. clinical geneticists, is the point of reference for ordering and returning laboratory tests: "It is expected that the clinician will contextualize these findings to the clinical circumstances (e.g., the nature of ongoing clinical problems, knowledge of personal and family history, patient preferences), and the provider and patient will participate in a shared decision-making process regarding the return of results. This is similar to the shared decision-making that is undertaken by patients and physicians: whenever complex medical testing is contemplated, patients are informed that data generated from such tests can reveal unexpected results". A strong concern is expressed on direct-to-consumer testing, where patients directly receive results from laboratories, but "Patients who seek out their laboratory test results independent of their healthcare providers have made their own choice about learning these results".

[30] See A. TOWNSEND, F. ROUSSEAU, J. FRIEDMAN, S. ADAM, Z. LOHN, P. BIRCH, *Autonomy and the patient's 'not to know' in clinical whole-genomic sequencing*, in *European Journal of Human Genetics*, 22, 2014, 6-11, questioning constructively the obligation to return IFs because of paternalism, predicating upon "the physician as gatekeeper" model, instead of the "patient as partner" or "patient as gatekeeper" models. They broadly rely on autonomy in Andorno sense, following in this paper, arguing that a chance must be offered in all pre-test and post-test counselling and the choice cannot be presumed, but must be activated by the patient. The position is based on a focus group study, whose participants strongly supported the right to decline information: "Individuals may have valid reasons for not knowing results. The "objective" clinical utility of knowledge may have very different meaning "subjectively" for patient regarding its relevance and manageability for them; all things considered, they simply may make an informed choice not to have the information disclosed to them. The impact that all genetic information has on the individual, including psychological, social and financial consequences or harms, needs to be reconsidered by clinicians at all stages of disclosure". In contrast, for further critics to "the right not to know" as an autonomy based right, because the right to refuse medical treatment is doubtful and with the aim to protect vulnerable people from the risk to decline medically

ISSN 2284-4503

confirms this solution and is stated as follows: An additional *modification to the original policy included offering an option to opt out of receiving SFs for individuals undergoing clinical genomic sequencing (GS).* This revision was due, in part, to results from a survey of ACMG members in which more than 80% of respondents supported an option for patients undergoing GS to decline SF analysis following appropriate counselling. In conclusion, for U.S. *Guidelines* now informed consent is necessary and reporting of secondary findings is optional.

Similarly, the issue has been deeply discussed during the French SFMPP works as well, and many doubts have arisen. The French working groups considered that genomic consent rules did not adequately fit with the disclosure of secondary findings from genome sequencing, nether under Art. L. 1111-4 *Code de la santé*, inspired by Art. 5 Oviedo Convention, nor as the informed consent under the *Loi Bioètique*. Art. 5 Oviedo Convention, in particular, may not be suitable with SFs because there is no "intervention", neither "act", nor "treatment". Moreover, there is no well-established, but only possible information. Again, predictability is one of the main problems. The disease is uncertain, but possible. It depends not only on the genome, but also on lifestyle, environment or other external factors. The quantity and quality of information have also been questioned because the lack of knowledge could depend on the scarcity of data, but even their large amount can be a problem, making their interpretation unclear and doubtful. Therefore, informed consent, as provided in general for genomic testing, may be unsuitable *per se* for the communication of secondary findings. The working groups questioned, in the end, the practical possibility of a right to know or not to know, because the object of knowledge is too uncertain and undefinable.

Ethically, four alternatives were discussed concerning the degree of disclosure, assuming autonomy in the meaning of making a life lived according to each own project: first, to be always informed (*être prévenu systématiquement*), expression of the principle of absolute autonomy, i.e. the right to know everything (*droit de tout connaître*); second, to be informed of relevant information (*être prévenu avec les informations pertinentes*), mirroring the principle of controlled autonomy depending on the relevance of data; third, not to be informed, unless the relevant information is strongly validated or clinical assistance is very recommended, mirroring again the principle of controlled autonomy, but focusing on a significant loss of chance; fourth, not to be informed at all. During the works, it has been specified that all alternatives imply a certain degree of paternalism, either in the obligation to know, as the only way to make choices and worthily organise life, or in completely avoiding knowledge, assuming that the patient could not understand the choice or to bear the pain. Finally, spoiling the end of the story, French *Guidelines* opt for a model of partial disclosure, admitting "the right not to know" of the patient as a central feature and adopting a new formula called "double consent", which dissociates the announcement of primary findings from SFs, as we will illustrate later.[31]

significant information, B.B. BERKMAN, S.C. HULL, L.G. BIESECKER, *Scrutinizing the Right Not to Know*, in *The American Journal of Bioethics*, 15, 7, 2015, 17-19.

[31] See the presentation *Consentement éclairé et aspects éthiques* of Ms. Michèle Anahory, lawyer, during the works of SFMMP working groups, available at https://www.sfmpp.org/2019/01/31/recommandations-de-la-sfmpp-sur-les-donnees-secondaires/ until November 2019. In January 2021, videos and power points were unavailable anymore. All mistakes reporting the contents are in my responsibility.

The Italian Bioethics Committee also referred to "the right not to know", and the report on IFs deeply discussed its rationale relying on the results of a Canadian Symposium held in Spring 2014.[32] The Committee identified three main meanings of autonomy as the source of the right to decline information: first, the protection from interferences of others in the most personal choices, requesting policies that enforce individual liberties, including "the right not to know" health data; second, autonomy as control by a rational agent of all circumstances of life, to make responsible and organised choices, and here there is no room for "the right not to know", lack of information preventing any rational decision,[33] third, "veridicity" in Hans Jonas words in the seventies, facing biomedical innovations, i.e. the right to find his own way of life and to shape the future. This last position supports a moral right to ignorance about the future, for example, for genetic diseases where no treatment is available, to remain free in determining the sense of self. However, these different theoretical models, in the Committee's opinion, did not help find a concrete solution if the patient had not expressed any will about being informed or not. Hence, the Committee referred to another theoretical scheme, Laurie's approach to privacy, which we will deeply analyse in the next paragraph. Finally, the Italian Committee, as we mentioned before, recognised centrality to self-determination, assigning priority to pre-test and post-test counselling with support of psychological expertise, and stating the existence of the right to choose whether to be informed and on which pathogenic results, actionable and non-actionable.[34]

Collectively, the U.S. *Guidelines* recommend the reporting as secondary findings of known or expected pathogenic variants and do not recommend reporting variants of uncertain significance or associated with disorders not currently amenable to intervention. The focus is mainly on SFs related to monogenic disorders for which there is a higher likelihood of causing the disease, i.e. evidence of clinical utility. Since 2015, informed consent is necessary, and reporting SFs is optional.

Similarly, French *Guidelines* of 2018 suggest reporting only pathogenic variations and clearly assure an opt-in opt-out option to patient, adopting the procedural approach of "double consent". In this way, for the French too, the knowledge of secondary findings is limited to variations where validated screening or prevention strategies are available, is optional, even if recommended, and it depends on the will of patient to know or not to know.

The Italian Bioethics Committee suggested that laboratories should store secondary findings, including non-known variations; centrality should be given to self-determination of the patient

[32] *From the Right to Know to the Right not to Know*, Special Issue in *Journal of Law, Medicine and Ethics*, 42, 1, 2014, *Special Issue,* 1-6.

[33] Report *Gestione degli "incidental findings"*, cit., 16, referring to J. HARRIS, K. KEYWOOD, *Ignorance, Information and Autonomy*, in *Theoretical Medicine*, 22, 5, 2001, 415-436.

[34] Report *Gestione degli "incidental findings"*, cit., 18. There is a sort of contradiction in the Report. In the previous part, concerning the different rationales of autonomy, the Committee observed that the interest and/or right not to know should be protected to the greatest extension. Later, it is said that being not absolute, the right can be limited, depending on factual circumstances, to protect other interests, that the Committee do not explicitly mention, in a case-by-case evaluation. In that context, said the Committee, it is not possible to completely avoid discretionary because *it is in the last resort to clinicians, or researchers, with the help of consultants, decide to 'exceptions', relying on the kind of information at stake*. Overall, while great relevance is given to the patient's choice by letting him to decide the kind of pathogenic variations to be known, the final decision is left to practitioners without any clear guideline.

ISSN 2284-4503

through consent, leaving him the choice to select which variations to know, pathogenic with clear clinical utility or pathogenic without available treatment. The difference from U.S. and French conclusions is consistent, involving non-actionable pathogenic variations.[35] In contrast, all recommendations refuse reporting and communication of unknown variations.

6. The right not to know: Legal basis and conceptual objections

The overruling of U.S. *Guidelines* from a mandatory disclosure of secondary findings concerning pathogenic variations to an optional choice, from 2013 to 2015, is a clear symptom of the very sensitivity of the issue and the complexity of the interests involved. The same impression comes from the French debate on the suitability of traditional informed consent because of the uncertainty of information itself. For the patient, now all guidelines clearly refer to the so-called right not to know. Despite the incertitude and the intense debate, "the right not to know" in health matters has a clear legal basis in international law. It dates to 1997 in both the UNESCO Declaration on the Human Genome and Human Rights, Art. 5.c, and in the Oviedo Convention on Human Right and Biomedicine, Art. 10.2.[36] The Oviedo Additional Protocol concerning Genetic Testing for Health Purpose, entered into force recently, on 1 July 2018, protects the wish of a person undergoing a genetic test not to be informed.[37] Under both the Oviedo Convention and its Protocol, the right to know or not to know is part of the right to respect private life. Also, a right to decline information is provided by many domestic legislations, and recently by Italian law on consent and non-resuscitation orders too, no. 219 of 2017.[38] Prior to this law, as we mentioned, rules were provided for under the Italian Data Protection Authority authorisation, with the specific task of regulating the processing of genetic data.

[35] *Gestione degli "incidental findings" nelle indagini genomiche con le nuove piattaforme tecnologiche*, cit., 18.

[36] "2. Everyone is entitled to know any information collected about his or her health. However, the wishes of individuals not to be so informed shall be observed. 3. In exceptional cases, restrictions may be placed by law on the exercise of the rights contained in paragraph 2 in the interests of the patient". The right not to know is recognised also in international clinical guideline, the *World Medical Association Declaration on the Rights of the Patient* of 1981, revised in 1995, Art. 7.d: "the patient has the right not to be informed on his/her explicit request, unless required for the protection of another person's life", and the *World Health Organisation Guidelines on Ethical Issues in Medical Genetics and the Provision of Genetic Services* of 1997, Table 7: "the wish of individual and families not to know genetic information, including test results, should be respected, except in testing of new born babies or children for treatable conditions".

[37] "2. Everyone undergoing a genetic test is entitled to know any information collected about his or her health derived from this test. The conclusions drawn from the test shall be accessible to the person concerned in a comprehensible form. 3. The wish of a person not to be informed shall be respected. 4. In exceptional cases, restrictions may be placed by law on the exercise of the rights contained in paragraphs 2 and 3 above in the interests of the person concerned".

[38] Art. 3 It. law no. 219 of 2017: "Ogni persona ha il diritto di conoscere le proprie condizioni di salute e di essere informata in modo completo, aggiornato e a lei comprensibile riguardo alla diagnosi, alla prognosi, ai benefici e ai rischi degli accertamenti diagnostici e dei trattamenti sanitari indicati, nonché riguardo alle possibili alternative e alle conseguenze dell'eventuale rifiuto del trattamento sanitario e dell'accertamento diagnostico o della rinuncia ai medesimi. Può rifiutare in tutto o in parte di ricevere le informazioni ovvero indicare i familiari o una persona di sua fiducia incaricati di riceverle e di esprimere il consenso in sua vece se il paziente lo vuole. Il rifiuto o la rinuncia alle informazioni e l'eventuale indicazione di un incaricato sono registrati nella cartella clinica e nel fascicolo sanitario elettronico […]".

For what is relevant here, this authorisation referred to secondary discoveries, stipulating that the information resulting from a genetic testing to be provided to the data subject included among the results the "unexpected news" and stating that the data subject was required to declare whether or not he wanted to know the results, including the unexpected one.[39] Differently, Italian law no. 219 of 2017 does not explicitly concern genetic testing but gives a general framework on consent and the substantive values it embodies. Perhaps terms like "informed consent", "diagnostic assessment", or "treatment", under this law, could not be suitable for genetic information, thereby raising the same concerns of the French debate resumed before. But we assume that the general framework provided by this law, giving centrality to the trusty clinical-patient relation, to dignity and self-determination of the patient and decisional autonomy and expertise of the practitioner, is broad enough to apply to genetic testing too. Moreover, we consider that IFs returning policies move rapidly, depending on increasing evidence-based data both on predictability of information and on psychological impact, so that, at this time, a more specific legislation may not be appropriate, while an adjustment based on clinical guidelines regularly updated, and hopefully standardised across Europe, seems desirable.

Despite these legal references, as we have seen, the clinical, ethical, and legal debate on IFs concentrated on the acceptability of a "not to know" pretension. Whole-genome sequencing has dramatically broadened the scope of available information.[40] Nevertheless, the general lines of the debate about the *rationale* of the right not to know have not significantly changed. Therefore, it seems useful to illustrate them.

The relation between the right to know or not to know and genetic information has been explored since the 2000s.[41] Literature specified that "the right not to know" one's genetic status raised several objections. Preliminarily, this right goes strongly against the trend of claiming, affirming and widening the right *to know* health information by patients, which has been fundamental to re-define the modern doctor-patient relationship. More generally, politically and philosophically, the right to remain in ignorance seems contrary to the desire to know, as a feature that distinguishes humans from other animals, and to the capability of knowledge as a prerequisite for decision, that is, at the foundation of legal rights since the Enlightenment.

Further, if we leave aside, for a moment, the issue of health information, from a constitutional perspective the right *to know* has been questioned by American law scholars since the 1960s as a

[39] See the DPA general authorisation no. 8 of 2016, cit., par. 5, lett. b) on unexpected results; par. 6, second sentence, on informed consent: "the person concerned is required to state whether or not he or she wishes to know the results of the examination or research, including any unexpected information concerning him or her, if it represents a concrete and direct benefit for the person concerned in terms of treatment or prevention or awareness of reproductive choices". The regulation is remained the same under the ongoing DPA authorisation no. 146 of 2019, par. 4.3 on *Information*, including unexpected news among the attainable results, par. 4.5, *sub* 3, on *Consent*, par. 4.6. on *Communication and dissemination of data*, under which: "the results of genetic testing must be communicated to the person concerned also in accordance with his or her declaration of willingness to know or not to know such events and, where necessary, together with appropriate genetic counselling" (our translation from Italian).

[40] B.M. KNOPPERS, *From the Right to Know to the Right Not to Know*, in *Journal of Law, Medicine and Ethics*, special issue 42, 1, 2014, 1-10, 6, who argues that the question now is not whether to return results, but how much information should be returned.

[41] For general references, R. CHADWICK, M. LEVITT, D. SHICKLE (eds.), *The Right to Know and the Right Not to Know. Genetic Privacy and Responsibility*, Cambridge, 2014 (second edition revised of the first dating 1997).

basic principle for democracy, referring it to the freedom of press and the public to acquire and share information on government and the exercise of public powers. Certainly, the context highly differs, but it is to say how *knowledge* is perceived as a stronghold, perhaps the most important, for democracy in Western societies.[42] Notably, even in that context, the right to know, as part of the liberty of expression, also covered a personal intimate position, essential to personal fulfilment. It is a significant method for seeking the truth, or at least for seeking the better answer, as it has been said.[43] In addition, in that context "the right not to know" emerged as the other side of the right to know, particularly in the meaning of not to be forced to receive communication. However, concurrently, it was relevant in narrower cases, much less significant for democracy than the right to know ones.[44]

Overall, knowledge seems to be the essence of human life. This is perfectly described in the famous words that the Italian poet Dante gave to Ulisse: "Fatti non foste a viver come bruti ma per seguir virtute e canoscenza"[45] [Consider your origin: you were not born to live like brutes, but to follow virtues and knowledge]. However, going back to health information, there are other objections to "the right not to know". It would undermine the relationship between doctor and patient, hindering the duty to disclose health information to the patient and returning to the paternalistic approach by which doctors are restricted to tell the truth, to supposedly protect the patient from harm. Moreover, "the right not to know" would be in contrast to the values of solidarity and responsibility, preventing the possibility of giving vital information to family members, in opposition to the relational nature of genetic information with relatives.[46]

[42] See for example in the U.S. debate, T. I. EMERSON, *Legal Foundations of the Right to Know*, in *Washington University Law Quarterly*, 1, 1976, 1-24, lecture delivered on March 3, 1976, during the Symposium *The First Amendment and the Right to Know*, discussing a constitutional theory and workable operating rules to anchor the right to know, an emerging constitutional right, in the U.S. Constitution. The A. opens the paper quoting a letter from James Madison to W.T. Barry, August 4, 1822, in *9 Writings of James Madison 103* (G. Hurst ed. 1910): "A popular government, without popular information or the means of acquiring it, is but a prologue to a farce or a tragedy; or perhaps both. Knowledge will forever govern ignorance. And people who mean to be their own governors, must arm themselves with the power knowledge gives".

[43] T.I. EMERSON, *op. cit.*, 2, 4: "Moreover, the right to know serves the same function in our society as the right to communicate. It is essential to personal self-fulfilment. It is a significant method for seeking the truth, or at least for seeking the better answer. It is necessary for collective decision-making in a democratic society. And it is vital as a mechanism for effectuating social change without resort to violence or undue coercion". Personal fulfilment is considered an important aspect because "our society moves further and further towards conformity and depersonalization".

[44] See T.I. EMERSON, *op. cit.*, 22-23, for whom the right not to know is a limit of the right to know, but much less meaningful than another opposite right, the right to privacy. While privacy needs the application of complex balancing theories, the right not to know, in U.S. Supreme Court case law at that time, refers to "simple" cases on the refusal to listen to music and broadcasting during bus travels, or to receive mails from anonymous senders. In general, concluded the A., the conflict is between the right not to know and the right to communicate, rather than the right to know.

[45] Dante Inferno, Canto ventiseiesimo, vv. 112-120.

[46] There would be also a practical impossibility of the right not to know, and a logical contradiction, because "in the very process of asking "Do you want to know whether you are at risk…?", the geneticist has already made the essence of the information known", in D.C. WERTZ, J.C. FLETCHER, *Privacy and Disclosure in Medical Genetics Examined in an Ethics of Care*, in *Bioethics*, 5, 3, 1991, 212-232, 221. The argument is fascinating, but seems

7. Theoretical frameworks between autonomy and privacy

Despite the referred objections, as we have seen, the right to refuse health information has a specific legal basis in international law, and in domestic law too, including now in Italian law. Different rationales of the right have been proposed to describe its nature and substance. We limit our analysis to two theoretical frameworks, which have had a great echo in the debate: "the right not to know" as an expression of autonomy and as an expression of privacy. The first thesis has been argued by Roberto Andorno. Against the argument that ignorance affects autonomy, preventing any choice and control,[47] Andorno argues that "the right not to know" is part of autonomy itself, understood as self-determination, mirroring the basic bioethical principle for which everyone should be free to make choices regarding health information. It is part of autonomy, without any paternalism, because the decision is in the patient's hands and does not come from abroad.[48] Autonomy is presented as the immediate source of the right, but the substantive interest protected in the end is psychological integrity, through the possibility to avoid the harmful effects of genetic information.[49] In Andorno's description, this right relies on an active approach from two perspectives. First, the opportunity of a choice is given to the patient. Second, the expression of will from the patient, being an explicit choice, is required[50]. Unlike, the privacy framework excludes the disclosure of information even if the patient has not explicitly expressed his or her will. This seems to be the main difference between the two frameworks, as we will see now.

The second framework, supported by Graeme Laurie, argues that "the right not to know" is not a right in itself, but an interference to privacy. While the framing paradigm of autonomy is choice, the framing paradigm of privacy is non-interference. More deeply, for Laurie, the ethical and legal basis of "the right not to know" is rooted in a "psychological spatial privacy, to the aim of safeguarding "one's own sense of the self".[51] The point is that spatial privacy should protect the patient even if no explicit choice has been made to assure that unsolicited revelations are prevented.

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invalid, because prior explanation of the possibility to find something does not say anything about the final output; so the effectiveness of the right relies, as always happens, on when and how information is given.

[47] For example, J. HARRIS, K. KEYWOOD, *Ignorance, Information and Autonomy*, cit, 415-436.

[48] R. ANDORNO, *The right not to know: An autonomy based approach*, in *Journal of Medical Ethics*, 30, 2004, 435-440, spec. 436, who refers this meaning of autonomy also to German legal literature, as part of the "right to informational self-determination" ["Recht auf informationelle Selbstbestimmung"].

[49] R. ANDORNO, cit., 436, recalling the oldest principle of medical clinics "first, do not harm" – "Primus non nocere," and the modern "principle of non-maleficence," including patient's psychological integrity.

[50] R. ANDORNO, cit., relies also on normative indications: The *European Convention on Human Rights and Biomedicine*, Art. 10.2, the UNESCO *Universal Declaration on the Human Genome and Human Rights*, Art. 5.c, as well as the WMA *Declaration on the Right of the Patient*, Art. 7.d, and the WHO *Guidelines on Ethical Issues in Medical Genetics and the Provision of Genetic Services*, Table 7, all require an explicit choice to the functioning of the right not to know.

[51] See G. LAURIE, *In defence of ignorance; genetic information and the right not to know*, in *European Journal of Health Law*, 6, 1999, 119-132; ID, *Protecting and promoting privacy in an uncertain world: Further defences of ignorance and the right not to know*, *ivi*, 7, 2000, 185-191; ID, *Challenging medical-legal norms. The role of autonomy, confidentiality, and privacy in protecting individual and familial group rights in genetic information*, in *Journal of Legal Medicine*, 22, 2001, 1-54; ID, *Genetic privacy. A Challenge to medico-legal norms*, Cambridge, 2000, 259, references made by R. ANDORNO, *op. cit.*, notes 30-36. See also ID, *Privacy and the right not to know:*

Collectively, the autonomy and privacy framework both protect the same substantive position, psychological integrity, and both admit that autonomy and privacy often overlap. Also, as Andorno himself noted, unsolicited information directly *interferes* with psychological integrity, passing by the choice on knowledge. However, the two theoretical frameworks differ in differentiating between general rule and exceptions. Under the privacy approach, non-interference is the rule, so that information becomes the exception. However, this effect, in Andorno's opinion, subverts the clinician's duty of disclosure principle. In fact, under the autonomy framework, the rule remains that the patient has the right to know his or her health status and that an alternative should be offered to him or her, if to have information or to remain in ignorance.[52] Non-information as a rule breaks this pattern. Overall, for the autonomy approach, "the right not to know" cannot be presumed but should be "activated" by the explicit will of the person.

Alternatively, it is true that the language of privacy has resonance with the ways that Western legal systems frame the interests involved.[53] Laurie recalls the "penumbra" definition of privacy in the U.S. Supreme Court case law on abortion and reproductive choices,[54] and, in Europe, the Strasbourg Court case law on Art. 8, par. 1, ECHR concerning respect for private and family life . But, the theoretical framework of Laurie's spatial privacy differs in reality from the ECHR interpretation. It is not the case here to deeply analyse the case law on 8 ECHR. It is enough to say that respect for private life under Art. 8 ECHR covers, depending on the circumstances of the case, personal autonomy, physical and psychological integrity, individual's psychological wellbeing and dignity.[55]

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a plea for conceptual clarity, in R. CHADWICK, M. LEVITT, D. SHICKLE (eds.), *The Right to Know and the Right Not to Know. Genetic Privacy and Responsibility*, cit., 38-51.

[52] R. ANDORNO, *op. cit.*, stresses that to determine which right should prevail will depend on the circumstances of each case, but law and ethics need rules to operate coherently. The right to know too, since its origin, had a positive shape. See T. I. EMERSON, cit., 2, for whom the right to know focuses on the affirmative aspects of the first amendment and the system of freedom of expression, the negative right to be free of government interferences being partial, meaning that the right is entitled to support by legislation or other affirmative government action.

[53] G. LAURIE, *Recognizing the Right Not to Know: Conceptual, Professional, and Legal Implications*, in *Journal of Law, Medicine and Ethics*, special issue 42, 1, 2014, 53-63, spec. 57.

[54] It is well known that privacy has different meanings, depending on each normative background and legal tradition. In a very general manner, at the origin U.S. privacy referred to a spatial dimension: it was intended to protect a *zone* of privacy within which the individual is protected against any intrusion from any rule or practise, public or private. Actually, the U.S. Supreme Court ruled on the constitutional foundation of consent under the *due process* clause of XIV Amendment on the grounds of a common law rule rooted in history and traditions, which considers forced medication as a battery. See R.R. FADEN, T.L. BEAUCHAMP, *A History and Theory of Informed Consent*, New York, Oxford, 1986, 120 ff., and, in Italian literature, C. CASONATO, *Introduzione al biodiritto*, Torino, 3rd ed., 2012, 160 ff. To question the right not to know, B.B. BERKMAN, S.C. HULL, L.G. BIESECKER, *Scrutinizing the Right Not to Know*, cit., 17, argue that the *Cruzan v. Director* case, 1990, concerning hydration and nutrition, related to bodily integrity, and cannot be extended to psychological integrity, falling outside the scope the XIV Amendment.

[55] See for all P. VAN DIJK, F. VAN HOOF, A. VAN RIJN, L. ZWAAK, *Theory and Practise of the European Convention on Human Rights*, 5th ed., Cambridge, Antwerp, Portland, 2018, 667-734. A recent summary of the case law is available in the Guide on Article 8 of the European Convention on Human Rights, dated 31.8.2020, at https://www.echr.coe.int/documents/guide_art_8_eng.pdf. The case law is divided in three categories: (i) a person's physical, psychological, or moral integrity, (ii) his privacy and (iii) his identity and autonomy. Under the first one, the rights protected include, among others, wellbeing and dignity (*Beizaras and Levickas v. Lithuania*,

Now, the Laurie's notion of space and preemptive non-interference fits with the original meaning of privacy, as undue home invasion from unlawful house searches or other police activities. It entails a negative obligation of abstention by public powers. However, the evolution of ECHR privacy, even concerning personal and intimate aspects of personality, has given increasingly importance to the so-called positive obligations.[56] Applicants claim a violation of eight ECHRs because of omissions by national authorities. Their quest is for an intervention of State at different levels, legislative, administrative or in practise, to put in place conditions to effectively develop their intimate expectations. It is also true that the opposition between negative and positive obligations is a false one, because negative obligations can easily be restated as positive obligations, and vice versa, and all rights can just as readily be described as having correlative obligations that are both positive and negative.[57] Anyway, the concept of positive obligation emphasises that standing is not enough for public authorities to respect rights.

Thus, in our opinion, Laurie's approach seems less convincing for two reasons. First, it recalls the ancient model of privacy as a duty of abstention from undue invasions but overshadows the positive duty of intervention implied in "the right not to know". For the good intention to avoid unsolicited information, it overlooks the complexity of positive and negative behaviours which are required by this right. Second, the "spatial privacy approach" seems to place the patient in a previous condition of isolation, supposing a pre-emptive opposition to intervention and, in some way, leaving him alone. Intervention as an assumed intrusion recalls an authoritarian, vertical model, while the doctor-patient relationship developed through years tends to a horizontal model, inspired by mutual trust. Further, if an unsolicited information is given, or a problem arises about sharing information or not,

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§117), personality development (*Von Hannover v. Germany* (no. 2) [GC], §95), physical and psychological integrity (*Söderman v. Sweden*, [GC], §80), relations with other human beings (*Couderc and Hachette Filipacchi Associés v. France* [GC], §83), the protection of personal data (*M.L. and W.W. v. Germany*, §87) and a person's image (*Reklos and Davourlis v. Greece*, §38). The ECHR ruled for the first time that the notion of private life covered the physical and moral integrity of the person in *X and Y v. the Netherlands*, 26 March 1985, §22, concerning the sexual assault of a mentally disabled sixteen-year old girl and the absence of criminal law provisions to provide her with effective and practical protection. Since this first case, the Court has held that the authorities' *positive obligations*—in some cases under Articles 2 or 3 ECHR*,* and in other instances under Article 8 taken alone or in combination with Article 3—may include a *duty to maintain and apply in practice an adequate legal framework affording protection*.

[56] See for all A.R. MOWBRAY, *The Development of Positive Obligations under the European Convention on Human Rights by the European Court of Human Rights*, London, 2004; M. KLATT, *Positive Obligations under the European Convention on Human Rights*, in *Zeitschrift für ausländisches öfentliches Recht und Völkerrecht = Heidelberg Journal of International Law*, 71, 4, 2011, 691-718, discussing positive and negative obligations and the principle of proportionality under Alexy theories. The main reasons why the negative wording of ECHR has been rephrased in positive terms are the overcome of the distinction between first generation, civil and political rights, and second generation, social rights, a pure passive approach to human rights being insufficient due to the complexity of our society, and indivisibility emerging under day-by-day situations; the need for effectiveness, especially after the reform of 1998 on individual applications; the main critics to this evolution rely on the lack of democratic accountability of the ECtHR, in. B. DICKINSON, *Positive Obligations and the European Court of Human Rights*, in *Northern Ireland Legal Quarterly*, *Special Issue*, 61, 3, 2010, 203-208, collecting the acts of a workshop on positive obligations in ECHR case law held in the Human Rights Centre of the School of Law at Queen's University Belfast, 24 March 2010.

[57] B. DICKINSON, *op. cit.*, 203.

probably a movement which breaks individual "separateness" has already happened and maybe it has been caused by the patient himself, because he or she has already made access to healthcare. Therefore, the question is how public authorities should manage the situation to let the patient, whose separateness has already been infringed, enjoy his or her right to refuse information. Overall, the spatial privacy approach, as mere abstention, contrary to the ECtHR case law, underestimates all conditions, tasks and duties required to make a right effective, which is at the core of Art. 8 ECHR interpretation.

Another debate on "the right not to know" concerns the effectiveness of freedom and the possibility of coercion. Briefly, criticism of the autonomy approach could be formulated considering that people are not always free to decide according to their real interests, because various forms of coercion, in a more or less explicit way, may interfere and press their will. We will see now that the effectiveness of freedom occupies the debate on negative or positive liberties.

8. Constitutional background between negative and positive liberty

It is well known that BioLaw shows a strong trans-nationality due to the cross-communication of scientists and the growing importance of international instruments, legally binding and non-binding. Scientific research is naturally cross-border, and its permeability implies a dilution of the distance between different constitutional cultures and a weakening of borders between common law and civil law systems. However, concurrently, there are certain "constitutional features" that depend on history, tradition, doctrine and practise. An attempt is made to analyse whether different approaches to set the right to know or not to know secondary findings could be justified by different constitutional cultures. The second part of this paper addresses this issue.

The relatively fast shifting of U.S. *Guidelines* on secondary findings from mandatory to optional disclosure, from 2013 to 2015, and the recognition of the right not to know, seem to put greater emphasis on autonomy of the patient as the only relevant principle at stake. As a first impression, this interpretation could rely on the primacy of personal liberty in the U.S. constitutional tradition, mistrusting government and public authorities' intervention affecting individual choices. This pattern comes after the origins of American society, where the smallholders refused positive interventions from authorities, finding a consensus on the core values of individual capacity, property and limitation of powers.[58] However, the strong emphasis which the first version of U.S. *Recommendations* put on the fiduciary duty of care and the clinician's obligation to prevent harm, to justify a mandatory disclosure, shows that this first impression is false.

Also, scholars' in-depth analysis has demonstrated that American constitutional tradition cannot be reduced to liberty as an isolated right from different perspectives.[59] First, American tradition is

[58] On the role of property as a model to describe personal rights, see later in the text.

[59] A. DI MARTINO, *La doppia dimensione dei diritti fondamentali*, in V. BALDINI (ed.), *Cos'è un diritto fondamentale*, Atti del Convegno Annuale di Cassino, 10–11 giugno 2016, Napoli, 2017, 123-158, spec. 136-142, and extensively in *Gruppodipisa.it*, no. 2 of 2016, who compares German, American and Italian constitutional backgrounds on rights and duties, using as a starting point the relation between individual dimension and institutional dimension, i.e. the institutional and organisational measures giving substance to rights, where the term "dimension" is taken from German constitutional interpretation. On Republican liberty see also I. CARTER,

pervaded by republicanism, which refers to common good, political participation and protection of liberties through the rule of law. Second, the U.S. Supreme Court case law, while discovering fundamental rights through the due process clause of the XIV Amendment, often refers to liberty and justice or, in more recent cases, ruled on freedoms that require a positive intervention of the State, for example, on the same-sex couple marriage.[60] Then, to give place to autonomy and personal development, public authorities must engage and adopt organisational measures. The same "positive" background is involved in the debate on privacy, if understood not only as a duty of abstention, but also as a duty of protection, to create and assure a place for freedom of choice despite different social and economic conditions. Third, different American constitutional theories associate rights and autonomy to responsibility, considering the promotional role played by the State.[61] Finally, concerning social rights, it has been demonstrated that they cannot be differentiated from civil rights regarding the costs and engagement of public authorities because even civil rights protection needs public expence and organisational measures.

This agrees with the American debate on the negative or positive meaning of liberty. The debate became stronger between social and political philosophers after the famous distinction defended by Isaiah Berlin in 1950s.[62] It has been demonstrated that many aspects of negative and positive liberty overlap, so that the distinction is not theoretically clear, but remains useful in a descriptive sense, at least to map different theories of liberty. Negative liberty, here, means the absence of obstacles or constraints, and usually refers to the absence of voluntary, intentional actions of others. Positive liberty is the possibility of acting, having control of one's life and realisation of one's purposes. Thus, positive liberty can be understood as self-realisation or self-determination. The concept gives relevance not only to external voluntary interventions of others, but also to internal barriers which could influence one's behaviour, for example ignorance, or fear, admitting interventions of the State to make the individual effectively free, interventions not normally allowed by a liberal conception.[63] As we said, the distinction has a descriptive meaning more than a prescriptive one. Libertarian and egalitarian in many cases overlap, giving different meanings to the constraint of freedom, where libertarians count as a restriction of freedom those brought about intentionally, being subject to the arbitrary will of another, and egalitarians endorse a broader conception of constraints as unintended obstacles, including economic forces, social conditions, poverty, instruction or other factors which

Positive and Negative Liberty, in *Stanford Encyclopedia of Philosophy*, in *plato.stanford.edu*, first published on February 27, 2003; last revision August 2, 2016, 5.

[60] A. DI MARTINO, quoted above.

[61] Compare the literature concerning a duty of protection of State, in A. DI MARTINO, cit., 28, who observes that a general category as the *Schutzpflichten* lacks in American tradition, but many Authors found a duty of protection principle rooted in the common law and translated into States' Constitutions.

[62] I. BERLIN, *Two Concepts of Liberty*, in I. BERLIN, *Four Essays on Liberty*, London, 1969, new edition in 2002.

[63] For a full discussion of Berlin's theory and scholars' different positions, I. CARTER, *Positive and Negative Liberty*, cit., devoting an extensive analysis to G. McCallum's "one concept of liberty" theory, in 1967, following F. Oppenheim studies in the 1950s., for whom the dichotomy between "freedom from" and "freedom to" is a false one and always refer to a triadic relation between three things, an agent, certain preventing conditions, and certain doings or becoming from agent, so that each theorist could be considered a supporter of the negative or positive one depending on the viewpoint. In sum "it is conceptually and historically misleading to divide theorists into two camps, a negative liberal one and a positive non-liberal one".

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are external, but also the presence of abilities, of capabilities, both influencing the internal sense of self and self-realisation.[64] These nuances lead us to a similar coexistence between negative and positive sides of liberty in the Italian constitutional framework.

9. Consent and the right of self-determination in the Italian constitutional framework

In the French legal system, free and informed consent is considered an expression of human dignity.[65] In Italian system, consent is usually described as a component of the right to self-determination.[66] This right is not explicitly mentioned under the Italian Constitution. Nevertheless, since the 1990s, the Italian Constitutional Court (ItCC) held that self-determination is a constitutional right. It could be useful to describe briefly how this right has been woven into Italian basic norms. The story of self-determination is like the current of a river, born as part of its natural flow, and then perceived as a single stream. Its origin is peculiar both because of the sources and the meaning. Starting from the sources, self-determination has always been strictly related to consent. Initially, consent was required by deontological sources, literature and justice, but not by law. In this way, rules for practise and a case-by-case approach lay the foundations of consent. As we have seen, only in 2017 has a general law on consent entered into force, turning the case-law principles into a legislative written text.

Regarding the meaning, self-determination was discovered by the ItCC as part of the right to health protected by Art. 32 It. Const. The Italian constitutional right to health is a very complex figure. It could be described, as the Italian constitutional scholars say, like a figure with different dimensions.[67] It comprehends liberties and duties, individual rights and general interests. Briefly, the constitutional right to health means freedom to choose if and how to seek treatments and, at the opposite, freedom not to seek treatment and to refuse them. Concurrently, health entails a typical social right, the right to access healthcare and to benefit from medical treatment. In other systems, the right to health could be framed differently. For example, it has been observed that the EU Charter of Fundamental Rights properly guarantees the freedom side and the social side under different articles.[68] Nevertheless, despite the different formulation, these two dimensions, the freedom to

[64] On types and sources of constraints in liberal and egalitarian theories and their nuances, and on the ambiguity of term "external" as the source of obstacles, I. CARTER, cit., 8, 9.

[65] Fr. Conseil Constitutionnel, 24 July 1994: "*le devoir d'information est un principe à valeur constitutionnelle et trouve son fondement dans l'exigence du respect du principe constitutionnel de la sauvegarde de la dignité de la personne humaine*".

[66] For a complete bibliography on informed consent, M. TOMASI, *Genetica e Costituzione*, cit., 38-39, note 82.

[67] Constitutional rights are described within dimensions in P. RIDOLA, *Diritti fondamentali. Un'introduzione*, Torino, 2006.

[68] A. CARMINATI, *Libertà di cura e autonomia del medico. Profili costituzionali*, Bari, 2018, 55-56, referring to Art. 3.2, *Right to the integrity of the person*, Title I, *Dignity*, for the right to consent: "1. Everyone has the right to respect for his or her physical and mental integrity. 2. In the fields of medicine and biology, the following must be respected in particular: (a) the free and informed consent of the person concerned, according to the procedures laid down by law […]", in which consent is part of the integrity of the body; Art. 35, *Healthcare*: "Everyone has the right of access to preventive healthcare and the right to benefit from medical treatment

choose treatments and the right to obtain them, are inevitably interconnected. The freedom dimension may be void if medical treatments are inaccessible, for example, because they are expensive. Liberty could be formally affirmed, but it is denied if choices are unaffordable.[69] Then, in the Italian constitutional framework, social rights to healthcare and healthcare organisations in general are strictly connected to the freedom dimension. A clear symptom of this interconnection lies in the wording of Art. 32 It. Const., which guarantees free medical care to the indigent. Also, under Art. 32 It. Const. health is a general interest and entails the obligation to behave under the law to achieve collective health. A typical example of health as a collective interest are mandatory vaccinations.[70]

Under this framework, ItCC found the basis for the right to self-determination. It has been discovered as a current in the health river, and it has become a single stream. Precisely, the right to self-determination has been derived from the second paragraph of Art. 32 It. Const. "No one may be obliged to undergo any health treatment except under the law". A consent to treatments should be necessary, to possibly refuse them.[71] Then, as a single stream, self-determination became an autonomous right in the ItCC case law.[72] This evolution had important consequences. While the meaning of heath extended moving from the organic functionality of the body to psychological

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under the conditions established by national laws and practices. A high level of human health protection shall be ensured in the definition and implementation of all the Union's policies and activities".

[69] B. PEZZINI, *Il diritto alla salute. Profili costituzionali*, in *Diritto e società*, 1985, 42-43.

[70] For a distinction between mandatory obligations and coactive obligations in ItCC case law concerning Art. 32 It. Const., on health, and Art. 13 It. Const., on personal liberty and integrity of the body, M. CARTABIA, *La giurisprudenza costituzionale relativa all'art. 32, secondo comma, della Costituzione italiana*, in *Quad. cost.*, no. 2, 2012, 455-465, spec. 456-460.

[71] E. ROSSI, *Profili giuridici del consenso informato: i fondamenti costituzionali e gli ambiti di applicazione*, in *Rivistaaic.it*, 2011, 6, who describes consent like a hinge between the negative dimension and the positive dimension of the right to health, i.e., the negative right to refuse actions of third parties causing damages and the positive right to ask for assistance and access to healthcare; in details, if mandatory medical treatments must be imposed only by law, all other treatments are voluntary, thus conditioned to consent. G.U. RESCIGNO, *Dal diritto di rifiutare un determinato trattamento sanitaria secondo l'art. 32, c. 2, Cost., al principio di autodeterminazione intorno alla propria vita*, in *Diritto pubblico*, 2008, 85 ff., spec. 91, insists on the fact that, to allow refusal, consent must be informed, i.e. imply a clear and qualified information. On the obligation of a qualified information as an essential attribute of consent, B. PEZZINI, *Il diritto alla salute. Profili costituzionali*, cit., 42-43. Following S. RODOTÀ, *Il nuovo habeas corpus: la persona costituzionalizzata e la sua autodeterminazione*, in S. RODOTÀ, M. TALLACCHINI (eds.), *Ambito e fonti del Biodiritto*, in S. RODOTÀ, P. ZATTI (eds.), *Trattato di Biodiritto*, Milano, 2010, 177, Art. 32.2 introduced a sort of new *habeas corpus*, parallel to the protection of physical integrity under Art. 13 It. Const. For these and more references, see A. CARMINATI, *Libertà di cura e autonomia del medico. Profili costituzionali*, cit., 24 ff., who emphasises that self-determination, and consent, is part of the right to physical integrity of our own body, protected by Art. 13 It. Const.

[72] Self-determination was born "in the shadow" of the right to health and the two proceeded together as travel companions for A. SANTOSUOSSO, *Autodeterminazione e diritto alla salute. Da compagni di viaggio a difficili conviventi*, in *Notizie di Politeia*, 1997, 27 ff. The leading cases are ItCC no. 438 of 2008 and no. 253 of 2009. In no. 438 of 2008 the independence of self-determination from health became clear. The autonomy of self-determination is evident in the case law of civil courts concerning medical treatments, where failure to collect consent, as an infringement of the right to self-determination, gives cause for compensation claims *per se*. On the relation between science and law in ItCC case law under a comparative perspective, deeply S. PENASA, *Presupposti scientifici delle leggi e giudizio di costituzionalità: spunti dal diritto comparato*, in M. D'AMICO, F. BIONDI (eds.), *La Corte costituzionale e i fatti: istruttoria ed effetti delle decisioni*, Milano, 2018, 255-269.

integrity, self-determination now comprehends personal aspects such as sexual identity, religious beliefs, different views on the beginning of life, education, self-development, the meaning of social relationships, and solidarity. Informed consent is in a way the bridge between the health river and the single stream of self-determination, giving protection to the whole complex of subjective aspirations and emotions that the physical dimension evokes.[73] Overall, a subjective perception of health emerged, parallel to its objective meaning.

10. *(continue)* The right not to know under Italian constitutional law

The question is whether the described constitutional framework fits with the challenges of secondary findings. Certainly, express reference made by Art. 32.2 It. Const. to the right to refuse treatments, combined with the constitutional right to self-determination, offers a secured, stable foundation to "the right not to know", as a negative expression of liberty. Italian Law no. 219 of 2017, under Art. 1, *Informed consent*, par. 3 states that every person "can refuse all or part of information or designate family members or a person of trust to receive them and give consent if this is the will of the patient".

However, the only anchor of self-determination may not be enough to set the issue. As the scholars have already demonstrated, genome information implies a strong relation between the individual and those who belong to the same family line. Genome holds messages that project the person back to the past and forward to the future horizontally and vertically.[74] So, the genome does not go along an individual dimension, but a relational dimension. This understanding has significant consequences in organising genomic information between the single person and the other family member. Then, a self-determination model that focuses on the individual as an isolated subject seems not to consider all the interests involved.

We could rephrase the problem from another constitutional viewpoint. If we select consent and the right to self-determination as the only parameter driving the right to know or not to know secondary findings, the risk is to approach the issue using a model like a very ancient freedom, the right to property. In this sense too, Italian constitutional evolution could be useful. Since the sixties of the past century the relation between each person and his or her body was questioned. The traditional paradigm, emerging from the liberal approach, considered this relation regarding property. The right of property was the model on which is still based, for example, Italian Civil Code of 1942, Art. 5, forbidding acts of disposition of his own body causing a permanent reduction of physical integrity, or opposing the law, public order or common decency. This model recalls dominion and all powers to use and abuse things within the limits of the law. After the entry into force of the Italian Constitution in 1948, many years passed before lawyers and judges finally changed paradigm. In the sixties of nineteen centuries, a joint reflection was undertaken between civil law and constitutional law scholars, and the result was a re-definition of this relation of person to his or her body regarding

[73] A. CARMINATI, *Libertà di cura e autonomia del medico. Profili costituzionali*, cit., 34.
[74] M. TOMASI, *Genetica e Costituzione*, cit., 14-15, 229 ff.

ISSN 2284-4503

responsibility and solidarity towards others.[75] Art. 2 It. Const. associates the inviolable rights of each human and the fundamental duties of solidarity. Art. 3 It. Const. recognises equal social dignity, and states that it is the duty of the Republic to remove those obstacles of an economic or social nature that constrain the freedom and equality of citizens, thereby impeding the full development of the human person and the effective participation in the political, economic and social organisation. Overall, freedom and equality both design the path of self-realisation and personal achievement in a relational context, associating freedom to solidarity and responsibility.[76] This seems to be the complete set of parameters fitting with genetics and the issue of secondary findings. In this sense, two interrelated positive aspects of "the right not to know" emerge, the first concerning the protection of family members, as we said, and the second dealing with the obligations to make the freedom of choice effective, regarding consent.

10. Right not to know and family members

We have seen in the first part of this paper that one of the main objections to "the right not to know" concerns responsibility towards others, first family members. Clarification should be made on the meaning of family. Certainly, family entails bloodline. Somehow genetics moves the concept of family counter current, focusing on bloody "natural" family, whose human relationship could lack affection and common will, while legal and constitutional protection has evolved concerning the social meaning of family. Definitively, due to the hereditary nature of genes, genetic information is relevant to relatives. So relatives could entail the same health risks and should have the possibility to access available health remedies, and to change way of life and behaviours as well.[77] Also, family comprehends, under constitutional meaning, spouses or partners, whose involvement could be opportune for example in reproductive choices, opening strong questions on the protection of future children.[78] Here we leave aside the situation of spouses and partners and we limit our analysis to the bloodline family. Both from a theoretical viewpoint and in practise, "the right not to know" cannot supersede the rights of family members.

In the autonomy approach mentioned above, the right to refuse knowledge is not absolute. As a relative right, it should be restricted when disclosure to the individual is necessary to avoid serious

[75] R. ROMBOLI, *La libertà di disporre del proprio corpo: art. 5*, in *Commentario del Codice civile Scialoja-Branca*, Roma, 1988; ID. (ed.), U. BRESCIA, A. PIZZORUSSO, *Atti di disposizione del proprio corpo*, Pisa, 2007, collecting the acts of the third joint seminar of the Doctoral School of Private Law and the Doctoral School of Constitutional Justice and Fundamental Rights. See more broadly, P. VERONESI, *Il corpo e la costituzione*, *Concretezza dei casi e astrattezza della norma*, Milano, 2007.

[76] M. TOMASI, *Genetica e Costituzione*, cit., 32, 48. Genetic information are truly intertwining lives in S. RODOTÀ, *La vita e le regole*, Milano, 2006, 71.

[77] Due to the relational nature of genetic information, some ethicists argue that the concept of genetic privacy is a contradiction in terms, in R. ANDORNO, *op. cit.*, 437, referring to A. SOMMERVILLE, *English v. Genetic Privacy: orthodoxy or oxymoron?* in *Journal of Medical Ethics*, 25, 1999, 144-150.

[78] On reproductive issue in bioethical perspective, for example N. JUTH, *The Right Not to Know and the Duty to Tell: The Case of Relatives*, in *Journal of Law, Medicine and Ethics*, 1, 2014, *Special Issue*, 38-52, and references to R. RHODES, *Genetic Links, Family Ties and Social Bonds: Rights and Responsibilities in the Face of Genetic Knowledge*, in *Journal of Medicine and Philosophy*, 23, 1, 1998, 10-30; T. TAKALA, M. HÄYRY, *Genetic Ignorance, Moral Obligations and Social Duties*, in *Journal of Medicine and Philosophy*, 25, 1, 2000, 107-113.

harm to family members, because some form of prevention or treatment is available.[79] Similarly, Italian Constitutional Court case law, since the first judgement in 1956, admitted that any right under Italian Constitution is not absolute, all rights and interests need to strike a fair balance.[80]

U.S. *Guidelines*, since the original version of 2013, have always considered patients together with family members. Variants with a higher likelihood of causing the disease are treated as potentially involving family from the pre-test counselling to the return, interpretation and communication of results. IFs are contextualised regarding personal and family history, in the awareness that further screening on relatives might be necessary. So, the interests of family members are a fully-fledged part of IFs' discovery and communication strategies. Nevertheless, recommendations have not specified any formula to involve relatives, nor a way to balance the right to know of relatives with "the right not to know" of the patient.

Under French *Guidelines*, reporting to patients' relatives about genetic discovery of a primary or secondary variant in actionable genes is assumed not only as moral responsibility but also, in terms of French law, as legal duty for the proband.[81] Because the disclosure to relatives can be particularly painful for families, even deleterious in some cases, French *Guidelines* state that how this information is given to relatives should be included in the informed consent to allow the patient a better understanding and consideration of the implications of results. An envisage solution is that the medical genetic team, if the patient so authorises, disclose the results to the family, thereby protecting as much as possible the patient's anonymity.

The Italian Report of the Bioethics Committee seems to recommend the option of disclosure to relatives as well. The pre-test counselling should give information about the consequences of eventual IFs on biological relationships and suggest that results could, and in some circumstance should, with proper precaution, be reported to family members in case of severe disease needing immediate interventions.[82]

All the examined recommendations, obviously, do not analyse different sets of relations between the patient and his family, cooperative or conflicting. At a deep insight, it could be said that any problem

[79] R. ANDORNO, *op. cit.*, 438.

[80] ItCC no. 1 of 1956, on limits as integral parts of rights; more recently, ItCC no. 85 of 2013, par. 9 *Cons. dir.*, the ILVA case, concerning the conflict between safeguarding job conditions and pollution, on the lack of absoluteness of constitutional rights and the need for a fair balance between health, environmental protection, jobs, and private economic activity.

[81] The *SFMPP Recommendations*, cit., 1736, quoting M. TOURAINE, *Décret no. 2013–527 relatif aux conditions de mise en œuvre de l'information de la parentèle dans le cadre d'un examen des caractéristiques génétiques à finalité médicale*, in santéMdasedl (ed.), 20 June 2013. M. TOMASI, *Genetica e Costituzione*, cit., 120-121, 125, observes that only French legislation concerning genetic data, without mentioning specifically SFs, provides a duty of communication to relatives, and the patient may ask the clinician to disclose information to remain anonymous. Comparatively, in most legal systems, regulation of the issue is not provided by legislation, but by soft laws and in the circle of practitioners' autonomy.

[82] Report *Gestione degli "incidental findings"*, cit., 18: "Le persone che si sottopongono al test devono, inoltre, essere informate che i risultati dell'analisi possono avere importanti implicazioni per i loro familiari e che, in questo caso, è opportuno, in certe circostanze doveroso, consentire a questi ultimi, con le dovute cautele e modalità, di venirne a conoscenza" and note no. 38, about a duty of communication: "Ad esempio nel caso di informazioni di malattie gravi per le quali è necessario mettere in atto subito misure preventive o terapeutiche".

concerning "the right not to know" does not arise when relatives could be autonomously involved in testing. In this situation, each adult individual will decide, independent of the others, about the opportunity to be tested and will eventually exercise "the right not to know". The main issue concerns situations where the patient has to discover genetic information because, if information is not made available, relatives unable to be personally investigated would suffer serious harm. In this case, a first way to facilitate communication goes with information and persuasion of the patient himself. It means explanation of the family nature of genomic information and encouragement of family involvement and discussion. This informal approach depends on counselling and consent, as we will see in the next paragraph.[83] If this approach is not useful, or not possible in the circumstances of the case, we argue that a mandatory *involvement* of family members is required, and that family members should be contacted, whenever possible, even against the will of the patient. As we said, family members should have the same opportunity for pre-test evaluation and information, and they are entitled to the same opt-in opt-out option. If their personal testing is not viable, certainly their need for protection may affect and endanger "the right not to know" of the patient.[84] This seems to be the main limit to the exercise of this right. In our opinion, this conclusion complies with the constitutional pattern described above, where self-determination goes with solidarity and responsibility.

11. The positive side of the right not to know: Consent

Positive obligation as a side of "the right not to know" relies essentially on consent. It is already well acquired that consent is not a single act, but a procedure. Its centrality being assumed, in recent years' literature specified its limits. Attempts are made to avoid consent as a mere ritual, a form to fill out to defend practitioners, rather than protect patients.[85] All the examined guidelines insist on the need and centrality of pre-test and post-test extensive counselling by the clinician who knows the

[83] This is a "responsibility-generating factor" approach for M. TOMASI, *Genetica e Costituzione*, cit., 119.

[84] This is in fact the solution provided for in the Italian DPA general authorisation. Compared to the first version in 2007, since 2011 the authorisation has been revised in the light of ongoing experience and experts' opinions to allow the processing and communication of genetic data indispensable to family members for health protection purposes without the consent of the data subject. The current text of the authorisation no. 146 of 2019, cit., provides that the results of a genetic test may be communicated to members of the same genetic line at their request when the data subject consented or when it is essential to avoid harm to their health, including reproductive risk, and the data subject's consent is not given or cannot be given due to unavailability (par. 4.6). Furthermore, par. 4.7 is expressly dedicated to the protection of the health of third parties and provides that if the consent of the data subject is not given or cannot be given due to physical impossibility, incapacity or unavailability, the processing of genetic data and their communication may be carried out when it is indispensable to enable the third party to make an informed reproductive choice or is justified by the need for preventive or therapeutic measures.

[85] G. CALABRESI, *Reflections on medical experimentation in humans*, in *Dedalus*, *Journal of the American Academy of Arts and Science*, 98, 1969, 405, considering that, as an academic, informed consent seemed to strike the best balance between different interests, while as a judge it was completely insufficient, because beyond the sheets of paper, people face showed they did not know what happened at all. [My apology if this quotation is incorrect; it was written in my notebook, and I was unable to check it anymore].

patient's physical conditions, history and expectations, with the help of geneticists or other consultants.[86]

The U.S. and Italian *Recommendations* do not set a specific procedure for the exercise of "the right not to know" SFs. In contrast, French *Guidelines* adopt a new scheme, the so-called double consent, which seems an interesting solution related to secondary findings.[87] The major point of this procedure is to respect the patient's desire to know or ignore results.

Briefly, double consent means that a first informed consent about the possibility of SFs is given during the initial medical procedure, motivating the primary genetic analysis. A second informed consent form is offered after the announcement of the primary findings, so that the patient could, with more autonomy, differentiate the issues and confirm or refuse access to this information. Information and informed consent must mention the medical impact of SFs on relatives. The clinical utility (regarding screening and prevention), for personal and family reasons, of a secondary discovery of a pathogenic variant within an actionable gene must be explained to the patient and counterbalanced by the arguments against this information. Because of the possibility to reconsider the given decision, SFs should be reported in a second report independent of the primary findings.

Two features characterise this formula. First, the dissociation of consent, and independence of the reports, concerning the primary investigation and secondary results. The need to dissociate the announcement of the primary findings from that of SFs was supported by patient associations to limit the psychological impact generated. Second, the role of time. Time is given, between the first and the second, confirmatory consent, to better understand and reflect. The main idea is a period of reflection, leaving the opportunity for patients to express their opinion again.[88]

Recent Italian Law no. 219 of 2017 on free and informed consent also emphasises the importance of time. Art. 8 states that the time of communication between the clinician and the patient represents the time of medical care ["Il tempo della comunicazione tra medico e paziente costituisce tempo di cura"]. And precisely in giving effectiveness to "time" comprises, in our opinion, the positive obligation, the active side of "the right not to know". Previous references to positive and negative

[86] The Italian Bioethics Committee's Report, for example, suggests the support of a psychologist in the pre-test counselling. Communication of IFs is for the clinician ordering the examination, due to the trust relation with the patient, but participation of other consultants is envisaged to better explain the results and their implications and to help plan next clinical steps. Outside the doctor-patient relationship, the Italian Committee insists properly on the need for extension of practitioners' competences in genetic counselling, including communication's abilities, and, more generally, for the implementation of school programmes and educational initiative on genetic issues, to improve knowledge of basic concepts of genetics, increase consciousness and the ability to form personal opinions. The U.S. *Guidelines* too mainly entrust the doctor-patient relationship with great flexibility.

[87] M. TOMASI, *Genetica e Costituzione*, cit., 254-287, analyses different models of consent, occurred over time to overcome the conditions of specificity and actuality required in traditional consent, like blanket consent, sectoral consent, multi-layered or dynamic consent, this latter suitable to communicate IFs.

[88] See in detail the diagram in Figure no. 3, *Recommended steps for reporting secondary findings to the patient*, in *SFMPP Recommendations*, cit., 1737. The second SFs consent, proposed during the reporting of primary findings, should also be offered again to patients who have stated in the first consent that they did not want to know the SFs. Thus, as for all patients, these patients will have the right to return to the decision after a period of reflection. Even if no formal pilot experiment of such a double consent was performed by the group, this agrees with the dynamic consent approach recently proposed to provide adaptive consent for research.

aspects of freedom are useful to argue, at the end, that the fulfilment of the right to know or not to know requires a specific, positive intervention to implement appropriate organisational measures. Literature on consent fosters dialogue and relational dimensions to overcome patients' constitutive asymmetry, scientific incompetence, and vulnerability. Reference is made to a dynamic process, scientifically comprehensive, devoid of technicalities, suited to the sociocultural conditions of the patient, and humanly sensitive.[89] Educational, financial and organisational investments are necessary to achieve those objectives. It has been specified that, under Italian Law on consent, the representation of time for information as part of the medical process remains a mere statement, because any budget has been allocated to implement it. Actually, there could not be time if structures and staff are not reinforced to perform ordinary tasks dealing with other duties.[90] Once again, "the right not to know" demonstrates that negative liberty is empty if it is not surrounded by measures and resources that make the opportunity of choice available, regarding understanding and affordability.

12. Conclusions: Double consent as a nudge

The paper overviewed the IFs issue from the clinicians' perspective, about actionability, and the patients' perspective, on "the right not to know", discussing two main theoretical rationales, autonomy and privacy, and moving from the US debate on positive and negative liberty to the French and Italian experiences. Concerning the pretension to refuse knowledge, the position of family members was analysed as the main limit to the individual right of choice. Furthermore, we argued that, to collect all interest, the negative exercise of the right not to know should rely on positive measures, shaping the procedure of consent.

As we said before, there is a certain degree of paternalism either in avoiding knowledge, assuming that patient cannot understand the choice or cannot bear the pain, or in the duty to inform and the obligation to know, as the only way to make choices and control circumstances of life. The double consent formula suggested by the French *Guidelines* in 2018 seems a good procedural attempt to strike a balance. During the works of the SFMMP, a short animation movie was promoted to explain to the public the issue of secondary findings and to disseminate recommendations.[91] The movie is centred on the patient, a little puppet, and accompanies him in a cosy atmosphere through hospital laboratories and counselling rooms to finally make the choice to have information or not. In a loyal and transparent way, it tends to suggest that knowledge of pathogenic variations could be useful to health. Can we consider this formula as a nudge, a small push, in the meaning of Thaler and

[89] See L. PALAZZANI, *Informed Consent, Experimentation and Emerging Ethical Problems*, in this *Review*, *Special Issue*, 1, 2019, *I-Consent. Improving the Guidelines for Informed Consent, Including Vulnerable Population, Under a Gender Perspective*, 11-22, spec. 16-18, where "time to reflect" is part of the suggested ethical requirements. I-Consent focuses on research, but many recommendations are suitable for clinical settings.

[90] See A. CARMINATI, *Libertà di cura e autonomia del medico. Profili costituzionali*, cit., 171.

[91] The animation movie, in French and English, is very useful to disseminate the issue, https://www.youtube.com/watch?v=Z1k3xN5rKvU. It is offered to patient and focuses on the opt-in opt-out chance.

ISSN 2284-4503

Sunstein?[92] A nudge is any aspect of the choice architecture that alters people's behaviour predictably without forbidding any options.[93] Nudges are everywhere, and push every minimal decision, but unthinkingly, as in advertisement. The authors explained that every day, we make decisions on topics ranging from school, education, meals and health, and we are susceptible to various biases. People make good choices in contexts in which they have experience, good information, and prompt feedback. They do less well in contexts in which they are inexperienced and poorly informed, and in which feedback is slow or infrequent. Choice architecture, both good and bad, is pervasive and unavoidable, and it greatly affects personal decisions. Therefore, the authors argued that public and private actors can design choice environments that make it easier for people to choose what is best for themselves, their families and society, without restricting freedom of decision. "Good nudges" could be "built" to drive people to appropriate decisions for themselves and their families. Particularly, people need nudges for decisions that are *difficult and rare*, for which they do not get prompt feedback, and when they have trouble translating aspects of the situation into terms that they can easily understand. Health is liable to be a choice environment. Doctors are presented as crucial choice architects, and with an understanding of how humans think, they could do far more to improve people's health. The authors' principal claim is that patients and doctors should be free to make their own agreements and that doctors' task is to help patients to map opportunities and to understand which option fits better with his personal wellbeing.[94] Thaler and Sunstein called themselves libertarian paternalists, suggesting that not only government, but employers, management boards, universities, clubs and single families could adopt measures to nudge members towards choices that will increase their wellbeing. They claimed that libertarian paternalism is not an oxymoron, because choice architects can preserve freedom of choice while also nudging people in directions that will improve their lives.[95]

This theory has been criticised exactly because of paternalism, and it is worth recalling that it moves from an economic framework, denying that consumer and client are rational agents. In fact, French *Guidelines* seem to be a real nudge in the sense of Thaler and Sunstein because they try to strike actionability, so high possibility of health measures, with the more intimate desire of patients about themselves, leaving them the final decision. Actually, concerning secondary findings, it is hard to find a settlement that is not, in some way, paternalistic, being constant the risk of applying one personal idea of rationality or wellbeing. In the end, a procedure divided into steps, bringing out different shades, based on time, allowing reflection, and preserving freedom, seems, in this complex pattern, a good solution to consider.

[92] R. THALER, C.R. SUNSTEIN, *Nudge, Improving Decisions about Health, Wealth and Happiness*, Yale University Press, 2008, *La spinta gentile. La nuova strategia per migliorare le nostre decisioni su denaro, salute, felicità*, trad. it. A. Oliveri, Milano, 2009 [VIII ed. 2018].
[93] *Ibid.*, 9.
[94] *Ibid.*, 102.
[95] *Ivi.*

Special issue

Newborn screening and informed consent in a constitutional perspective. The Italian "model" and some knots still to be untied

Caterina Di Costanzo*

ABSTRACT: Starting from variability analyses in newborn screening systems at international, European and regional levels, this paper focuses on the Italian "model" highlighting some critical issues in a constitutional perspective. The law n. 167 of 2016 and the ministerial decree of 13 October 2016 allowed to achieve uniformity in the national diagnostic offer but their mutual inconsistencies contributed to a regional variability in the management of informed consent/dissent and in the management of the biological material collected. The protection of fundamental rights at stake still requires a constitutionally oriented harmonisation and development of the rules governing the newborn screening national system.

KEYWORDS: Newborn screening; informed consent; informed dissent; constitutional principles; regional variability

SUMMARY: 1. Introduction – 2. Remarks about variability in the use of existing screening tests – 2.1. The variability of screening tests at international and European level – 2.2. The variability of screening tests at Italian regional level – 3. The Italian "model" and the rules on newborn screening in a constitutional perspective – 4. Regional practices after the rules of 2016 – 4.1. Regional practices on development of screening programmes and on management of informed consent and dissent to the procedure – 4.2. Regional practices on storage and use of biological material – 5. Some conclusive reflections.

1. Introduction

Neonatal screening is one of the most important secondary public preventive medicine programmes.

It falls within the secondary type of prevention which, unlike vaccines which are of the primary type and include all interventions intended to hinder the onset of diseases in the population and are aimed at so-called herd immunity, has the primary purpose of protecting the health of the newborn; the objectives of such screening are the early identification of those who are sick, preventing the onset and progression of the disease.

Screening is not a diagnostic assessment but a generalised diagnostic investigation strategy on a given population. It is therefore a predictive test that constitutes the initial moment of a diagnostic process which, after completing a further "confirmation test" (biochemical, enzymatic and/or molecular), allows the formulation of a certain diagnosis.

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ISSN 2284-4503

Most of the screening concerns the analysis of the genetic mutations of the newborn.
Diseases subject to newborn screening tests are rare congenital and inherited endocrine and metabolic disorders.[1]
The history of screening as a population test dates back to the early 1960s in the United States of America when biologist Robert Guthrie developed a simple and inexpensive bacterial inhibition test capable of identifying the most common aminoacid disease: phenylketonuria.
During the following decade, neonatal screening for congenital hypothyroidism and subsequently for cystic fibrosis (implemented with different methods) began both in the USA and in Europe, including Italy.[2]
In Italy, neonatal screening for phenylketonuria was first introduced in Italian Region Liguria in 1973 and afterwards was gradually widened to include the entire nation.
In another Italian Region, Tuscany, screening for phenylketonuria, hypothyroidism and cystic fibrosis was established in 1983, and this then became mandatory nationally in 1992, with law n. 104 of 5 February 1992 and the subsequent regulations for implementation.
In the 1990s, the development of analytical technology – tandem mass spectrometry – allowed the development of versatile, specific and sensitive analytical methods that made it possible to measure many biomarkers in a single and very rapid analysis.
This technology has made it possible to move from the concept of "one test – one disease" to that of "one test – many diseases", effectively revolutionizing the approach to newborn screening tests.
The criteria used date back to the 1960s, and these aim to identify the panel of diseases to be controlled.
The best known, defined by the World Health Organization, date back to 1968 and are known as the Wilson and Jungner criteria; these refer to both the characteristics of the disease (severity, frequency, possibility of dietary and/or pharmacological treatment able to improve quality and life expectancy) and the characteristics of the screening test (appropriateness, costs, acceptability by the population).[3]
Neonatal screening is normally carried out between 48 and 72 hours after birth and involves the collection of a few drops of blood. The main problems regarding newborn screening tests are not related to the invasiveness, which is very low, nor with the balance between risks and benefits of the

[1] Inherited Metabolic Disorders (IMDs), also called inborn errors of metabolism, constitute an important category of rare genetic diseases caused by the altered functioning of a specific metabolic pathway. They represent a heterogeneous group of over 700 different pathologies which, taken individually, are rare but together have a cumulative incidence ranging from 1 in 500 to 1 in 4,000 live births. From the point of view of public health, IMDs represent a group of pathologies with a considerable impact on the health of the person, the family and on society as a whole, as they are multi-systemic diseases that can cause irreversible damage to many organs and systems, responsible for early neonatal mortality and permanent psychic and neuromotor delays since childhood.

[2] See J.G. LOEBER, P. BURGARD, M.C. CORNEL, T. RIGTER, S.S. WEINREICH, K. RUPP, G.F. HOFFMANN, L. VITTOZZI, *Newborn screening programmes in Europe; arguments and efforts regarding harmonization. Part 1 - From blood spot to screening result*, in *Journal of inherited metabolic disease*, 35, 2012, 603-611; D.B. JR. BAILEY, *Early intervention and newborn screening parallel roads or divergent highways?*, in *Infants & young children*, January-March 2021.

[3] See J.M.G. WILSON, G. JUNGNER, *Principles and practice of screening for disease*, WHO, 1968.

act itself, since there are no risks to the physical well-being of the newborn; rather, the issues for concern are around the management of the autonomy profile and the self-determination of the legal representatives of the newborn in relation above all to the information that can be derived from the screening and future management of the data of the newborn.

2. Remarks about variability in the use of existing screening tests

There is extreme variability in this area at international, European, national and regional levels, since there is no international consensus on what should be included in the panel of diseases to be controlled; this is in spite of increased attempts to coordinate the screening programmes.[4] The fundamental question that emerges, having observed this divergence, is whether some countries or regions overdiagnose while others register a diagnostic supply deficit.

2.1. The variability of screening tests at international and European level

At international and European level, the diversity of the health systems is a relevant issue in reference to financing, insurance or the public system; in the identification of the primary recipients of the screening system (the newborn, the parents, the society); and in the interpretation of the concept of clinical utility (referring to possible treatment and cure of the disease or as a possibility for treatment and improvement of the quality of life and prognosis).[5]

Political decision makers in various European countries give different weight to the various factors involved in the screening processes, and the consideration given to the different evaluation possibilities of the screening systems is notably different.

A lack of harmonisation among European countries emerge in the provision of information about newborn screening to parents and emphasised the need for more comprehensive guidelines at the European level.[6]

The practices of access to screening are also differentiated,[7] including systems that refer to an opt-out clause, whereby the test is considered mandatory and presumes consent, on the basis that it provides protection in the best interests of the child, but nevertheless leaves open the possibility for parents or legal representatives to disagree and refuse it; to an opt-in model, which requires

[4] Cfr. the initiatives of the International society for neonatal screening (ISNS), https://isns-neoscreening.org/.

[5] The extensive screening is discussed and questioned in literature, because of the massive collection of data not necessarily related to immediate or actual clinical utility, so to the individual health of the patient. See M.S. Househ, B. Aldosari, A. Alanazi, A.W. Kushniruk, E.M. Borycki, *Big data, big problems: a healthcare perspective*, in *Studies in Health Technology and Informatics*, 238, 2017; Q.K. Fatt, A. Ramadas, *The usefulness and challenges of big data in healthcare*, in *Journal of Healthcare Communications*, 3, 2018.

[6] V. Franková, R.O. Driscoll, M.E. Jansen, J.G. Loeber, V. Kožich, J. Bonham, P. Borde, I. Brincat, D. Cheillan, E. Dekkers, R. Fingerhut, I.B. Kuš, P. Girginoudis, U. Groselj, D. Hougaard, M. Knapková, G. la Marca, I. Malniece, M.I. Nanu, U. Nennstiel, N. Olkhovych, M. Oltarzewski, R.D. Pettersen, G. Racz; K. Reinson, D. Salimbayeva, J. Songailiene, L. Vilarinho, M. Vogazianos, R.H. Zetterström, M. Zeyda, *Regulatory landscape of providing information on newborn screening to parents across Europe*, in *European Journal of Human Genetics*, 2020, 1-10.

[7] About this aspect see R. Brownsword, J. Wale, *In ordinary times, in extraordinary times: consent, newborn screening, genetics and pandemics*, in this issue.

informed consent to the procedure; and to hybrid models. Other discrepancies exist regarding the regulations around the retention of biological material beyond the time strictly necessary for carrying out the tests, and the possible uses to which the profiles can be put, including research not specifically linked to the screening.[8]

In addition to the number and type of diseases to be controlled, the profile relating to consideration of the clinical validity and clinical usefulness of the test, in particular the latter, is recorded and evaluated differently in the various countries.

Internationally, newborn screening programmes are active in more than 64 countries.[9] At European level, the wide variability in the use of screening has been highlighted starting from the final report of the survey on newborn screening[10] launched following the approval of the Council Recommendation on rare diseases of 2009,[11] and this has been outlined even more recently.[12]

The systems are significantly different in each country, and this difference affects the number and types of diseases to be controlled: for example, in Great Britain, the disease panel includes 9 pathologies; in Italy 47; in the Netherlands 34; in the Czech Republic 20; in Spain 7; in Ireland 6; and in France 5.[13]

It is evident that at European level, Italy offers the widest diagnostic in the screening sector.

2.2. The variability of screening tests at Italian regional level

At Italian regional level, analysing the experiences of three Italian Regions, such as Liguria, Emilia Romagna and Tuscany, some interesting insights emerge on the extent of variation in the screening offer.

As mentioned, Liguria was the first Region in Italy to introduce the screening test for phenylketonuria, in August 1973, with the regional law n. 31.[14]

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[8] Cfr. P. Burgard, M. Cornel, F. Di Filippo, G. Haege, G.F. Hoffmann, M. Lindner, J.G. Loeber, T. Rigter, K. Rupp, D. Taruscio, L. Vittozzi, S. Weinreich, *Short executive summary of the report on the practices of newborn screening for rare disorders implemented in Member States of the European Union, candidate, potential candidate and EFTA Countries*, October 2011; J. Kraszewski, T. Burke, S. Rosenbaum, *Legal issues in newborn screening: implications for public health practice and policy,* in *Public health reports*, 2006; B.M. Knoppers, D. Avard, K. Sénécal, *Newborn screening programmes: emerging biobanks?*, in *Norsk Epidemiologi*, 21, 2, 2012, 163-168.

[9] B.L. Therrel, C.D. Padilla, J.G. Loeber et al., *Current status of newborn screening worldwide: 2015*, in *Seminars in perinatology*, 39, 3, 2015, 171-187.

[10] P. Burgard, M. Cornell, F. Di Filippo et al., *Report on the practices of newborn screening for rare disorders implemented in Member States of the European Union, Candidate, Potential Candidate and EFTA Countries*, 2012, http://ec.europa.eu/chafea /documents/news/Report_NBS_Current_Practices_20120108_FINAL.pdf.

[11] European Council, *Council Recommendation of 8 June 2009 on an action in the field of rare diseases*, 2009.

[12] J.G. Loeber, *The European Union should actively stimulate and harmonise neonatal screening initiatives*, in *International journal of neonatal screening*, 4, 2018; B.L. Therrell, C.D. Padilla, J.G. Loeber, I. Khneisser, A. Saadallah, G.J.C. Borrajo, J. Adams, *Current status of newborn screening worldwide 2015*, cit.

[13] N. Meade, J. Spink, *Let's grasp this opportunity to examine the potential future of screening*, in *BioNews*, November 2019, https://www.bionews.org.uk/page_146203. For the Italian system see Servizio Studi della Camera dei Deputati, *I nuovi livelli essenziali di assistenza*, 12 January 2021, 7.

[14] Cfr. regional law n. 31 of 17 August 1973, *Regulations for the identification and treatment of phenylketonuric disease*.

Subsequently, with the implementation of regional law n. 26 of 8 September 1986, regional law n. 31 was abrogated, and screening for hypothyroidism was also introduced.[15]

The Ligurian system appears complex, because although there are no regional regulations, the screening tests have been carried out as a pilot project since 2005 as part of the regional newborn screening programme, ensuring coverage of all newborns in the Region.

The panel of controlled diseases with screening starting from 2005 was very large and was equivalent to that implemented in Tuscany. Indeed, about thirty diseases were screened as part of the mentioned pilot project coordinated by the Gaslini teaching hospital based in Genoa.

The screening system in Emilia Romagna was launched only in 2010.

With the regional regulation n. 107 of 1 February 2010, *Enlargement of screening for inherited metabolic disorders*, the number of diseases to be controlled was extended to 19 pathologies in addition to the three provided for as mandatory by the national law n. 104.[16]

In relation to this regional regulation a trial occurred before the administrative judge.

The decision of the Regional Administrative Court of Emilia Romagna of 17 December 2010 n. 8138 and the decision of the Council of State, the Italian Supreme Administrative Court, of 19 January 2012 n. 247 concerned the challenge of regulation n. 107 which determined the progression of the regional screening system.

The complaint to the Regional Administrative Court was promoted by a Patients' Association against the Emilia Romagna Region.

The main object of the plea was the annulment of the disputed part of the regulation in which the screening of hereditary metabolic diseases does not include all diagnosable diseases processed through the technology of tandem mass spectrometry; or, subordinately, in the part in which the early mass screening excludes a series of pathologies that are listed by the claimants.

The plea has been declared inadmissible because it required an integration of the regional provision through an action for annulment that could not lead to this effect.

Anyway, the Regional Administrative Court goes partially through the matter and states that the regional regulations did not violate, as alleged by claimants, the right to health of Emilian citizens because the provisions extended the execution of newborn screening well beyond the three pathologies provided nationally (to 19 pathologies in addition to the three declared mandatory by national law n. 104).

[15] Cfr. regional law n. 26 of 8 September 1986, *Regulations for the identification and treatment of hypothyroidism and phenylketonuria diseases*.

[16] The regional regulation n. 1898 of December 19, 2011, *Establishment of the hub and spoke network for hereditary metabolic diseases subject to newborn screening and organization of the path of global care of the pediatric patient*, and the regional regulation n. 365 of 27 March 2017, *First implementing measure in the context of territorial care of the Prime Minister's Decree of 12 January 2017 on the definition and updating of the essential levels of care pursuant to art. 1, paragraph 7, of the legislative decree 30 December 1992, n. 502* published in the Official Gazette n. 65 of 18 March 2017, constitute regulations with organizational functions of the regional screening system. The regional regulation n. 2260 of 27 December 2018 was approved in implementation of the law of 19 August 2016 n. 167 and the ministerial decree of 13 October 2016 relating to newborn screening for the early diagnosis of hereditary metabolic diseases and regional provisions on the subject.

The ruling of the Council of State two years later confirmed the first stage decision of the Regional Administrative Court, rejected the appeal, and focused on interesting aspects: the criteria for including and excluding diseases from the screening test.

The Council of State affirmed that in the absence of "universally shared international scientific criteria", the choice of diseases to be controlled is inspired by the fundamental principle of "diagnostic opportunity/usefulness" or, if preferred, "costs/benefits". In fact, it was not possible to manage an excessive number of false positives with the possible consequence of subjecting "clinically healthy subjects" to "inappropriate and continuous therapies" or of causing "anxiety for a long time to families". Even more difficult to manage was the risk of a single false negative.

The administrative judge concluded that "the choice whether or not to include certain pathologies in the mass screening (which are very rare by definition) involves very delicate profiles (for example because, for some of them, the intrinsic margin of error could cause more harm than good); hence the high degree of discretion removes the decision from judicial review competence, once it has been ascertained that it was adopted with reasonableness and thoughtfulness".[17]

Finally, Tuscany, more than other Regions, has a deep-rooted tradition of developing screening.

In Tuscany, from 1983, screening for phenylketonuria, congenital hypothyroidism, and subsequently for cystic fibrosis, was introduced; these screenings were subsequently made mandatory throughout the nation with article 6 of the law of February 5, 1992 n. 104.

The extended screening system began as a pilot project in 2002, and in 2004 it was launched throughout the Region with the specific regional regulation of 2 August, 2004.

Since 1 November 2004 with regional regulation n. 800 of 2004, screening in Tuscany has been extended to about 30 other metabolic diseases besides phenylketonuria, by means of mass tandem analysis.

The regional regulation n. 420 of 2018 concerning the *Tuscan Extended Neonatal Screening System: Update on the basis of the Decree of the Ministry of Health of 13/10/2016 and Law n. 167/2016 on the subject of diagnostic tests for the prevention and treatment of hereditary metabolic diseases* reaffirmed the function of the University Hospital Meyer, based in Florence, in coordinating the screening system, with the task of governing and monitoring the activities of the regional neonatal screening system as a whole. It confirmed that with the introduction of galactosemia following the ministerial decree of 13/10/2016, the panel of pathologies that in the Tuscany Region, currently subject to extended and mandatory neonatal screening, corresponds to the list in Annex A of the ministerial decree of 13/10/2016.[18]

[17] See ruling of the Council of State of 19 January 2012 n. 247.

[18] A further development occurs with the regional regulation n. 909 of 6 August 2018 *Extended neonatal screening for the early diagnosis of metabolic diseases and hereditary immunodeficiencies. Further development of the regional screening programme*. The Tuscan system also included other diseases that were not initially fostered in National rules and then inserted following by the 2019 budget law. It should be noted that from 1 January 2006 the Healthcare Authority n. 1 of the Umbria Region carries out the extended screening at the University Hospital Meyer and from 1 January 2010, according to the Memorandum of Understanding referred to in regional regulation n. 1277/2009, concerning *Regulation n. 236/2004 "Interregional framework agreement between the Tuscany Region and the Umbria Region for the management of healthcare mobility*", the neonatal screening activities have been extended to the entire territory of the Umbria Region according to a renewable three-year agreement.

3. The Italian "model" and the rules on newborn screening in a constitutional perspective

The Italian newborn screening "model" is the most developed system among those in Europe, considering that it currently has as its object a panel of 47 diseases to be controlled. The Italian system has developed since the 1990s and with the 2016 legislation – law n. 167 of 2016 and ministerial decree of 13 October 2016 – has substantially implemented the Tuscan diagnostic proposal that has represented the more developed regional model since the 1990s-2000s.

As mentioned, screenings for phenylketonuria, hypothyroidism and cystic fibrosis have been made mandatory at the national level starting from article 6, letter g, of the law 5 February 1992, n. 104 – *Framework law for assistance, social integration and the rights of disabled people* and subsequent implementing acts.[19]

Starting from the law n. 244 of 24 December 2007 (2008 Finance Law) important funding has been allocated (about 3 million euros) for the purchase of new analytical methods, based on "tandem mass spectrometry", to carry out expanded newborn screening for hereditary metabolic diseases, where there is scientific evidence that therapy is effective.

Law n. 147 of 2013, *Provisions for the preparation of the annual and multi-year budget of the State* (Stability Law 2014), in paragraph 229 of article 1 establishes: "[...] the experimental launch throughout the national territory, within the limit of 5 million euros, of neonatal screening for the early diagnosis of hereditary metabolic diseases, for whose therapy, pharmacological or dietary, there is scientific evidence of therapeutic efficacy or for which there is scientific evidence that an early diagnosis, in neonatal age, entails an advantage in terms of access to therapies in an advanced state of experimentation, including dietary ones".[20]

The same provision states that the Minister of Health should approve a ministerial decree, after consulting the Higher Institute of Health and the Permanent Conference for relations between the State, the Regions and the Autonomous Provinces of Trento and Bolzano, to define the list of pathologies on which the screening should be carried out and the procedures for its implementation.

A relevant change is undoubtedly marked by law n. 167 of 2016, *Provisions on mandatory neonatal diagnostic tests for the prevention and treatment of hereditary metabolic diseases*, which came into force on 15 September 2016, as it provides for the inclusion of extended neonatal screening (ENS) in the new Essential Levels of Care (ELC) so as to be able to guarantee access to ENS for all newborns nationwide.[21]

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[19] See the Prime Minister Decree of July 9, 1999 *Act of guidance and coordination for the regions and autonomous provinces of Trento and Bolzano in the matter of investigations for the early diagnosis of malformations and mandatory control for the identification and timely treatment of congenital hypothyroidism, phenylketonuria and cystic fibrosis*. See also the law n. 548 of 23 December 1993 *Rules for the prevention and treatment of cystic fibrosis*.

[20] Law n. 190 of 2014 (2015 stability law), in paragraph 167 of art. 1, then increased the National Health Fund by a further 5 million euros, starting from 2015, thus increasing the funds for extended neonatal screening (ENS) to 10 million euros a year.

[21] Law n. 167 of 2016 provides for the inclusion in the Essential Levels of Care (ELC) of mandatory neonatal screenings for the early diagnosis of hereditary metabolic diseases; it establishes the Coordination Center on neonatal screening at the Higher Institute of Health; it establishes that the Ministry of Health should prepare an operational protocol for the management of screening and for taking care of the sick; it assigns to the

On March 19, 2017, the date of enforcement of the Prime Ministerial Decree of January 12, 2017 which updated the ELC, the ENS passed from the experimental phase to full operation. In fact, in the provision of the new ELC, in article 38 paragraph 2 of the Prime Ministerial Decree of 12 January 2017, the inclusion of ENS was envisaged, referring to a ministerial decree for the list of diseases and the methods of implementation.

The decree of the Ministry of Health of 13 October 2016, *Provisions for the start of newborn screening for the early diagnosis of hereditary metabolic diseases* contains a series of indications concerning the list of pathologies covered by ENS: the information and consent procedures; the methods of collecting, sending and storing the blood spot; the organization of the newborn screening system, whether regional or interregional, to ensure the continuity of the entire ENS path from the first-level test, to the second-level test, to diagnostic confirmation; and taking charge of the confirmed positive cases in newborns.

However, there are important inconsistencies between law n. 167 of 2016 and the ministerial decree of 13 October 2016 which must be highlighted.

Law n. 167 establishes the inclusion in the ELC of neonatal screenings which are qualified as mandatory and therefore the tests become the responsibility of the National Health Service.[22]

While law n. 167 qualifies the screening as mandatory, the ministerial decree of 13 October 2016 contains in article 2 rules about information and consent for non-mandatory screenings[23], and Annex

National Agency for Regional Health Services the evaluation of the Health Technology Assessment (HTA) on newborn screening; and finally it provides for the method of financing the activity in question and establishes that from the entry into force of the Prime Minister's Decree that updates the ELC, the experimentation, started with law n. 147/2013, art. 1 paragraph 229, ceased. Subsequently, the 2019 budget law made important changes, extending the screening to genetic neuromuscular diseases, severe congenital immunodeficiencies, and lysosomal storage diseases.

[22] See art. 1 and art. 2 of law n. 167: art. 1 (Purpose): "This law aims to ensure the prevention of hereditary metabolic diseases, by including mandatory neonatal screening in the essential levels of care (ELC) [...]"; art. 2 (Scope of application): "The diagnostic tests as part of the mandatory screening referred to in art. 1 are carried out for hereditary metabolic diseases for which there is scientific evidence of therapeutic efficacy, pharmacological or dietary, or for which there is scientific evidence that an early diagnosis, in neonatal age, involves an advantage in terms of access to therapies in advanced state of experimentation, including dietary ones".

[23] Art. 2, titled "Information and consent", of the ministerial decree of 13 October 2016 is particularly interesting for our purposes and it reads as follows: "1. The ENS is carried out, after suitable information referred to in paragraph 2, provided to the interested parties by the professionals of the birth point. Where national or regional acts do not establish the obligation for the execution of the ENS, informed consent must be obtained for the execution of the ENS and for the processing of the personal data of the newborn, pursuant to art. 13 of the legislative decree 30 June 2003, n. 196, issued by natural parents or by the person exercising parental responsibility over the newborn. 2. The information, drawn up by the regions and autonomous provinces in an easily understandable language and translated into the languages most widely used in the area, must briefly and colloquially specify what the purposes and methods of the ENS are; the optional or mandatory nature of the screening procedure; the specific aims pursued (treatment and, if the ENS gives a positive result, genetic counselling); the methods of carrying out the test and the diseases tested; the achievable results, including any unexpected news known as a result of the differential diagnostics of the diseases referred to in attached table 3, which share the primary markers with those listed in table 1; the methods and times of storage of the samples; the scope of data communication, especially with reference to neonatal screening laboratories, clinical reference centres and the National Register of rare diseases, to which the data are

A to the ministerial decree presents a model of informed consent to be given to parents and legal representatives.[24]

Actually, the two fundamental acts of the Italian legislation on newborn screening seem to be mutually inconsistent.

From a strictly formal point of view, the ministerial decree of October 13 2016 refers in its foreword to law n. 167 but appears to be implementing paragraph 229 of article 1 of the Stability Law of 2014.[25]

From a substantial point of view, a series of critical issues emerge from a constitutional perspective.

First of all, by article 2 paragraph 1 of the ministerial decree of 2016, which reads: "[...] Where national or regional acts do not establish the obligation to execute the ENS, informed consent must be acquired for the execution of the ENS and the processing of personal data of the newborn […]", it could be understood that at the regional level, before the 2016 legislation, the screening tests provided could be considered mandatory. The first problem therefore concerns the possibility of qualifying a test as mandatory at regional level.

As known, art. 32, paragraph 2, of the Italian Constitution places a State law reserve when it requires a State law for the imposition of a mandatory health treatment that does not violate the limits of respect for the person.

On this point, a clear response comes from constitutional case law issued on the basis of the State legislation reserve contained in article 32, second paragraph, of the Italian Constitution on mandatory health treatments and on the basis of the division of legislative competences between the State and the Regions in the exclusive and concurrent matter of health guarantee contained respectively in second paragraph, letter m, of article 117 (determination of essential levels of care) and in third paragraph of article 117 (health protection), of the Italian Constitution. In decision n. 5 of 2018, the Italian Constitutional Court affirms that it is up to the State "to qualify a certain health treatment as mandatory, on the basis of the medical and scientific knowledge available".[26]

The discipline of mandatory treatments is straightforwardly assigned to the State legislative competence as it belongs to the determination of the fundamental principles concerning the matter of health protection.[27]

Since the criterion of the voluntary or mandatory nature of the treatments affects fundamental rights, such as the right to self-determination and the right to health that belong to the State

communicated through the regional registers. 3. The collection of informed consent, referred to in paragraph 1, must be carried out before carrying out the screening test, according to the contents of the model in Annex A to this decree, which is an example. It must contain the consent to carry out the screening, to the processing of data and to the storage of samples".

[24] As mentioned, in the first annex to the ministerial decree, a list of about 40 metabolic diseases is drawn up, which constitutes the most complete panel of diseases at European level, while in annex A of the decree an informed consent model is prepared which raises some issues.

[25] Art. 1, paragraph 229, of the law n. 147 of 27 December 2013 (Stability Law of 2014) referred to a ministerial regulation the definition of the list of diseases on which to carry out newborn screening.

[26] See the ruling of the Italian Constitutional Court n. 5 of 2018, paragraph n. 7.2.2 of the decision.

[27] With reference to vaccinations see the decision of the Italian Constitutional Court n. 137 of 2019 and n. 5 of 2018.

legislative competence, it can be assumed that a regional act cannot decide whether a treatment is mandatory.[28]

A second, relevant problem concerns the normative qualification of screening as mandatory or voluntary. While in article 1 of the law n. 167 of 2016 it is stated that neonatal screenings have become mandatory, in article 2 of the ministerial decree of 2016, a series of rules on information and informed consent are provided for *tamquam non esset*, except from a strictly formal point of view, the law n. 167. Looking at Annex A of the ministerial decree of 2016 and reading the model of informed consent reported there, it could be understood that the mandatory nature refers only to the three diseases provided for by the old law n. 104 of 1992, while everything else is framed as voluntary neonatal screening.

Leaving aside here the overview issue of the reserve contained in article 32 of the Italian Constitution as an absolute reserve of law, also reinforced by the provision of the respect for the human person,[29] or a relative reserve,[30] the contrast substantially existing between the law n. 167 of 2016 and the ministerial decree of October 13 2016 poses, *de facto*, some questions about the respect for the hierarchy of sources and highlights significant interpretative problems that have led to a great variability in clinical practice in the screening sector at regional level.

These problems require an in-depth study on the mandatory and voluntary nature of a test or a treatment.

As required by the Italian Constitution in article 32, second paragraph, only a State law, i.e. a primary source, can declare a treatment voluntary, even by not specifying anything on the point of obligation and therefore enhancing the constitutional principle of self-determination, or mandatory, when this is aimed not only at improving or preserving the conditions of health of those subjected to it, but also at guaranteeing the collective interest to health, since it is precisely this further proposal that justifies the suppression of individual self-determination.[31]

The definition of a treatment or a test as mandatory/voluntary is assigned to a State primary source as it belongs to the determination of the fundamental principles concerning the right to health.

In addition, the Italian Constitution provided for a reinforced legal reservation when it established that "The law cannot in any case violate the limits imposed by respect for the human person"[32] and

[28] See the decision of the Italian Constitutional Court n. 438 of 2008.

[29] On this position see P. BARILE, *Diritti dell'uomo e libertà fondamentali*, Bologna, 1984, p. 385; A. PACE, *La libertà di riunione nella Costituzione italiana*, Milano, 1967, 87 ff.; B. PEZZINI, *Il diritto alla salute: profili costituzionali*, in *Diritto e Società*, 1983, 28 ff.

[30] On this position see M. LUCIANI, *Il diritto costituzionale alla salute*, in *Diritto e Società*, 1980, 10; F. MODUGNO, *Trattamenti sanitari «non obbligatori» e Costituzione*, in *Diritto e Società*, 1982, 309; V. CRISAFULLI, *In tema di emotrasfusioni obbligatorie*, in *Diritto e Società*, 1984, 558; S.P. PANUNZIO, *Trattamenti sanitari obbligatori e Costituzione*, in *Diritto e Società*, 1979, 900; E. CAVASINO, *La flessibilità del diritto alla salute*, Napoli, 2012, 181.

[31] See the decisions of the Italian Constitutional Court n. 268 of 2017, n. 107 of 2012, n. 226 of 2000, n. 118 of 1996, n. 258 of 1994 and n. 307 of 1990.

[32] The Court affirmed in judgment n. 194 of 1996 that "blood sampling – now of ordinary administration in medical practice – does not harm the dignity or psyche of the person, just as it does not normally endanger his life, safety and health in any way (see decision n. 54 of 1986)". The limits of respect for the human person can be identified with the very low degree of invasiveness and its effects on the psycho-physical integrity of the person. The assessment also concerns the incisiveness that the test has on the freedom and self-determination of the person. With respect to this it is necessary to verify the possible provision of a compulsory treatment,

prevents such a qualification, related to the mandatory or voluntary nature of a test, being made from a secondary source, including, therefore by a ministerial decree.[33]

The constitutional case law on the matter is extensive and the relevant constitutional principles ensue from articles 2, 13, 32 of the Italian Constitution. The Italian Constitutional Court affirms that the balance between the individual's right to health and the interest of the community must take place in such a way that the right of the individual is not excessively affected except for temporary, minor and in any case tolerable consequences.

Over the years, the Court's case law has clarified the requisites necessary for the purposes of a legislative provision of an obligation of test or treatment. First of all, a justification of reasonable scientific efficacy at an epidemiological level is needed for the prevention of the disease in the subject undergoing test and for the management of a significant risk to public health and the legislative provision of measures to contain as much as possible the risks of adverse events.

Furthermore, on the basis of the solidarity duty that presides over the subject of the mandatory tests and treatments, compensation must be provided to compensate for any damage, even if not of a purely financial nature, suffered by the person who has undergone the treatment or test.[34]

In the case of newborns, there is a need for balance between the protection of the best interests of the child,[35] the exercise of parental responsibility, and the collective interest in preventing serious diseases whose management impacts on the community.

As stated, screening does not strictly represent a health treatment but constitutes an investigation strategy carried out in the context of secondary prevention.

We need to specify that the collective interest protected in the case of neonatal screening is of a different nature from the collective interest protected in the case of vaccines. While in the case of vaccines the collective interest is directly related to health, aimed at protecting both individuals and public health, in the case of screening the collective interest is not directly linked to protection of public health, but it does have an impact on health and social costs, considering the very high impact that metabolic diseases have on the person, the family, and the community because they are multi-systemic diseases that can cause irreversible damage to several organs and systems and are responsible for early neonatal mortality as well as permanent psychic and neuromotor delays from childhood.[36]

As we said before, massive screening program are disputed, but the possibility of preventing these inherited disorders or stopping their progression may undoubtedly correspond to an important collective interest of a socio-economic nature.

Similar examples that can be referenced in defining the contours of this collective interest and which also have an indirect impact on health, are the obligation to wear a helmet or seatbelt when riding or

which would require further, pursuant to art. 13 of the Italian Constitution, the legislative discipline of cases and methods of coercion and a provision of the judicial authority.

[33] It is necessary to specify here that once a national law has established that a treatment or a test is mandatory, a ministerial decree cannot decide otherwise as if the State law did not exist.

[34] See the decisions of the Italian Constitutional Court nn. 5 of 2018, 258 of 94, 307 of 90.

[35] On the relevance of protection of the best interests of the child for mandatory screening, see M. TOMASI, *Genetica e Costituzione. Esercizi di uguaglianza, solidarietà e responsabilità*, Napoli, 2019, 294 ff.

[36] See supra footnote n. 1.

driving vehicles. The obligation to use these safety measures does not have an immediate health purpose. Its function is to protect the individual and, from the collective point of view, is not aimed at protecting public health but at containing a risk, which can be statistically highlighted, and which is linked to significant economic and social effects on the national system and on the National Health Service.

As in the case of newborn screening, there is therefore an economic-functional link with respect to public health that is the basis of the imposition of the obligation that protects the collective interest and the health of the individual.[37] In this sense, it is possible to state that only a primary source, i. e. a State law, can define the boundaries of what should be mandatory and establish its effects.

4. Regional practices after the rules of 2016

As we mentioned before, paragraph 2 of article 117 of the Italian Constitution establishes that the "determination of essential levels of care" constitutes an exclusive legislative competence of the State, while paragraph 3 of art. 117 of the Italian Constitution includes the "health protection" competence among the concurrent competences. As part of this latter competence, it should be specified that the fundamental principles are provided for by the law of the State while the organizational rules are established at the regional level.[38]

On the basis of these constitutional norms, while the law of the State establishes which are the essential levels of care that must be guaranteed throughout the national territory (see the Prime Ministerial Decree of January 12, 2017), the Regions establish the procedures and organizational practices aimed at meeting the essential levels established. Within this constitutional framework of division of legislative powers between State and Regions, regional practices often differ and lead to very different organizational models.

In this context, the aforementioned inconsistency between the two 2016 acts on newborn screening has led to an increase in regional divergences in the ways of carrying out screening, managing informed consent and dissent, and managing the storage and the use of the biological material collected.

About the mentioned inconsistency of the two 2016 acts, we could deduce that according to law n. 167 extended newborn screening is mandatory and included in the ELC, while on the basis of the ministerial decree of 13 October 2016 the expression of consent concerns only the extended newborn screening which concerns diseases listed in the annex to the ministerial decree, while the three mandatory screenings remain those established by law n. 104 of 1992.

It must be said that there would be a need for a specific implementation of law n. 167 in order to overcome the critical issues envisaged. However, on the basis of the principle of conservation of legal acts, the interpreters carried out some operations of mutual adjustment and adaptation between the two acts. These operations were successful in some respects and not in others, as emerges from the analysis of regional practices subsequent to the 2016 rules. In fact, while on the one hand these norms, together with the decree updating the essential levels of care, made it possible to obtain

[37] See the decisions of the Italian Constitutional Court n. 180 del 1994 and ordinance n. 49 del 2009.

[38] Cfr. decisions of the Italian Constitutional Court n. 510 of 2002, n. 329 of 2003, n. 338 of 2003.

uniformity in the provision of the screening offer, the same uniformity cannot be traced in the management of informed consent and dissent. In addition, it seems evident that some aspects, such as the management of the conservation of the biological material collected and the use of the same for research purposes, require further analysis.

4.1. Regional practices with reference to development of screening programmes and to management of informed consent and dissent to the procedure

In order to highlight some features of regional practices on the subject, we will refer to the three reports, so far prepared by the Italian Higher Institute of Health and the Coordination Center on Neonatal Screening (CCSN), established by article 3 of the law n. 167 of 2016[39], in collaboration with the National Center for Rare Diseases, on monitoring the state of implementation of law n. 167 of 2106 and of the ministerial decree of 13 October 2016.[40]

In the first monitoring report on the state of implementation of law n. 167/2016 and the ministerial decree of 13 October 2016 on Extended Neonatal Screening (ENS) in Italy[41], the state of the art of the individual regional programmes of the ENS as of 30 June 2017 is described. The data highlighted that 18 Regions/Autonomous Provinces out of 20 Regions and two Autonomous Provinces on that date had started the ENS programme, while in 3 Regions the ENS system was still in the activation phase at the indicated date.[42]

The second monitoring report on the state of implementation of the 2016 rules provides an update on the evolution of the ENS system in the Regions, and documents the changes introduced on 30 September 2018 through a fact-finding survey conducted by the Coordination Center on Neonatal Screening (CCSN), with the aim of highlighting the new regional actions and strategies introduced between 30 June 2017 and 30 September 2018.[43] The state of the art on 30 September 2018 indicates that all the Regions/Autonomous Provinces, except Calabria, had started the ENS programmes. The ENS regional system in Calabria was, in fact, in the activation phase. Of the other Regions/Autonomous Provinces which had started the system, almost all covered the entire panel of metabolic diseases provided for in the ministerial decree of 13 October 2016.

[39] One of the tasks assigned to the CCSN (Article 3 of Law 167/2016) is to monitor and promote the maximum uniformity of application of newborn screening in Italy.

[40] We refer to, in chronological order, Higher Institute of Health, *Screening neonatale esteso nelle Regioni: monitoraggio dell'attuazione della Legge 167/2016 e del decreto ministeriale del 13 ottobre 2016. Stato dell'arte al 30 giugno 2017*, Rapporto Istisan 18/11; Higher Institute of Health, *Screening neonatale esteso in Italia: stato dell'arte al 30 settembre 2018*, 2019; Higher Institute of Health, *Programmi di screening neonatale esteso nelle Regioni e Province autonome in Italia. Stato dell'arte al 30 giugno 2019*, Rapporto Istisan, 20/18.

[41] Higher Institute of Health, *Screening neonatale esteso nelle Regioni: monitoraggio dell'attuazione della Legge 167/2016 e del decreto ministeriale del 13 ottobre 2016. Stato dell'arte al 30 giugno 2017*, Rapporto Istisan, 18/11.

[42] Not all Regions have defined the various levels of articulation of the ENS system, based on art. 4 of the ministerial decree of 13 October 2016. Furthermore, only 50% of the Regions have full coverage of the pathologies covered by the ENS.

[43] See Higher Institute of Health, *Screening neonatale esteso in Italia: stato dell'arte al 30 settembre 2018*, 2019.

In the Regions of Abruzzo, Campania, Friuli Venezia Giulia, Lombardy, and the Autonomous Provinces of Bolzano and Tuscany, new information sheets have been created to make parents and legal representatives aware of the purposes of ENS.

The Regions of Basilicata, Friuli Venezia Giulia, Molise, Umbria and Valle d'Aosta and the Autonomous Provinces of Bolzano and Trento have activated the ENS system in agreement with other Regions. Moreover, 10 Regions have issued new specific administrative acts for the ENS Regional System (Abruzzo, Basilicata, Campania, Emilia Romagna, Lazio, Lombardy, Piedmont, Valle d'Aosta, Tuscany and Veneto).[44]

In conclusion, in Italy, the ENS for the prevention, early diagnosis and treatment of Inherited Metabolic Disorders (IMDs), despite the inconsistencies existing between the law n. 167 of 2016 and the ministerial decree of 13 October 2016, passed from an experimental phase to a mandatory offer phase on all newborns in the national territory.

The third report of the Higher Institute of Health on screening concerns a survey that specifically provides information on regional practices relating to the management of informed consent and dissent to the procedure.[45]

As part of the fact-finding survey, the Regions/Autonomous Provinces were asked whether, for the execution of the ENS, informed consent was required from parents and legal representatives of the newborn.[46]

The results are interesting: 7 Regions and Autonomous Provinces (Campania, Lombardy, and the Autonomous Provinces of Trento, Puglia, Tuscany, Umbria, Valle d'Aosta) declared that they did not require informed consent; among the remaining Regions and Autonomous Provinces, the request for informed consent takes place in 4 Regions only for diseases not provided for in Annex A of the ministerial decree of 13 October 2016;[47] in 7 Regions the collection of informed consent occurs for provided and not provided diseases in Annex A of the ministerial decree of 13 October 2016;[48] and in 3 Regions only for the diseases listed in Annex A of the ministerial decree of 13 October 2016.[49]

Regarding the consent and dissent to the execution of ENS, the answers provided by the Regions/Autonomous Provinces seem to highlight a situation of chaos relating to the issue. In fact, half of the respondents require informed consent for "provided" or "not provided" diseases in Annex A of the ministerial decree of 13 October 2016 and do not require the systematic collection of dissent to the execution of ENS.

In conclusion, the rules of 2016 have essentially made it possible to move from a hybrid model – in which, with respect to three diseases at national level, on the basis of law n. 104 of 1992, a

[44] See Higher Institute of Health, *Screening neonatale esteso in Italia: stato dell'arte al 30 settembre 2018*, 2019, 4 ff.

[45] See Higher Institute of Health, *Programmi di screening neonatale esteso nelle Regioni e Province autonome in Italia. Stato dell'arte al 30 giugno 2019*, 4-9.

[46] See Rapporto Istisan, 20/18, *Programmi di screening neonatale esteso nelle Regioni e Province autonome in Italia. Stato dell'arte al 30 giugno 2019*, 7 ff.

[47] They are Friuli Venezia Giulia, Autonomous Province of Bolzano, Sicily, Veneto (Laboratory of Verona).

[48] They are Abruzzo, Emilia-Romagna, Lazio, Molise, Piemonte, Sardegna, Veneto (Laboratory of Padua).

[49] They are Basilicata, Liguria e Marche. It is useful to specify that Calabria did not provide the requested information. See Higher Institute of Health, *Programmi di screening neonatale esteso nelle Regioni e Province autonome in Italia. Stato dell'arte al 30 giugno 2019*, 7.

mandatory nature was envisaged, while at the regional level the wider diagnostic offer was based on a voluntary basis of the investigations – to an opting out model allowing for the implementation of uniformity of practice in the field of tests and procedures.

This uniformity achieved in terms of the diagnostic offer does not correspond to a similar uniformity in terms of regional practices as regards the awareness of the effects of the mandatory tests and the organizational management of informed consent and dissent, and in terms of the collection of informed consent for aspects not envisaged as mandatory by the rules of 2016.

The information that emerges from the fact-finding survey on the organizational management of consent and dissent highlights some unresolved issues.

Given the absence of uniform management at regional level, the Higher Institute of Health provides some recommendations on how to manage informed consent and dissent.[50]

On the basis of the analyses developed before in paragraph n. 3, and considered the inconsistencies between the two normative acts of 2016, we can affirm that the recommendations of the Higher Institute of Health are consistent with the constitutional oriented reading of the rules on the mandatory/voluntary nature of treatments and tests and on informed consent/dissent that we have tried to outline *supra*.

In fact, the Higher Institute of Health specifies that it should be necessary to ensure that informed consent would be acquired in the case of the execution of ENS for diseases not provided for by the ministerial decree of 13/10/2016 or for diseases other than those mentioned by other laws that make screening mandatory.

In this way, the principles established by the Italian Constitution in article 32, second paragraph, according to which only a State law, i.e. a primary source, can declare a treatment or a test voluntary or mandatory, are fully respected.

About the dissent, the Higher Institute of Health clarifies that the parental couple can express their objection to the execution of the mandatory ENS. However, it is necessary that the Birth Center transcribes this dissent on the information form, affixing the date and simultaneously acquiring the signature of those exercising parental responsibility and the signature of the healthcare professionals involved. The information form containing the dissent must be included in the medical record of the newborn and in the case of a home birth in the obstetric record.

4.2. Regional practices on storage and use of biological material

From another point of view, the Higher Institute of Health checked also the uniformity of regional storage and use practices. Since the cardboard containing the blood spot collected for the extended neonatal screening of metabolic diseases contains biological material of significant value, both in terms of public health and research, it must be stored in ways that allow respect for the fundamental rights at stake (e.g. family autonomy and self-determination, privacy, etc.)

From the data reported in the third report of the Higher Institute of Health and related to regional practices, we see that there is extreme variability in terms of time and methods of storage of the

[50] See Higher Institute of Health, *Programmi di screening neonatale esteso nelle Regioni e Province autonome in Italia. Stato dell'arte al 30 giugno 2019*, 8-9.

card containing the blood spot.[51] From these data it is clear that there is a lack of process norms to regulate the methods of conservation and use of the residual material on the cardboard containing the blood spot, not only for diagnostic and medical legal purposes but also for research purposes, to ensure uniformity of the storage conditions and the methods of use at national level.

In the absence of any explicit regulatory provision on this point, it can be assumed that the biological material of the newborn, the drops of blood on the card, can be stored only for the time strictly necessary for carrying out the tests related to screening.

Beyond this time, the biological material should be removed or, if possible, the data of the newborn should be anonymised.[52]

If the biological material is kept for research reasons beyond the time necessary to carry out the tests, it can be concluded that these aspects could be the subject of informed consent, both regarding the conservation profile and the research uses, and for concerns about the processing of data and its codification and pseudonymisation.

The point has an obvious relevance and it is necessary to distinguish the uses that are made of the biological material in relation to the screening tests and the uses that can be made in relation to future research not related to the screening tests. The management models for conservation and future research on biological material, based on a minimisation of data processing (codification/pseudonymisation/anonymisation), and the authorisation of the competent ethics committee, may be various and may require adequate information to be given to the parents and legal representatives; the collection of the parents' informed consent may, in turn, have a variable scope and could *in abstracto* include alternatives, ranging widely from specific consent to partially restricted consent, tiered consent up to broad consent.[53]

5. Some conclusive reflections

There is no doubt that the newborn screening programme developed in Italy represents the most advanced "model" in Europe – from a specific viewpoint, i.e. the panel of the diseases to be checked – and that the research has achieved significant objectives in this area. The constant scientific research work on the subject portends a progressive expansion of screening programmes. The

[51] See Higher Institute of Health, *Programmi di screening neonatale esteso nelle Regioni e Province autonome in Italia. Stato dell'arte al 30 giugno 2019*, 31-32.

[52] The real possibility of anomymization is questioned in literature. See F.K. Dankar, A. Ptitsyn, S.K. Dankar, *The development of large-scale de-identified biomedical databases in the age of genomics-principles and challenges*, in *Human Genomics*, 10 April 2018.

[53] On the possible modelling of informed consent for scientific research cfr. C. Grady, L. Eckstein, B. Berkman, D. Brock, R. Cook-Deegan, S.M. Fullerton, H. Greely, M.G. Hansson, S. Hull, S. Kim, B. Lo, R. Pentz, L. Rodriguez, C. Weil, B.S. Wilfond, D. Wendler, *Broad consent for research with biological samples: workshop conclusions*, in *American journal of bioethics*, 15, 9, 2015, 34-42; R.B. Mikkelsen, M. Gjerris, G. Waldemar et al., *Broad consent for biobanks is best – provided it is also deep*, in *BMC Medical Ethics*, 20, 2019; J. Murphy, J. Scott, D. Kaufman, G. Geller, L. LeRoy, K. Hudson, *Public perspectives on informed consent for biobanking*, in *American journal of public health*, 99, 12, 2009, 2128-2134; E. Salvaterra, L. Lecchi, S. Giovanelli, B. Butti, M.T. Bardella, P.A. Bertazzi, S. Bosari, G. Coggi, D.A. Coviello, F. Lalatta, M. Moggio, M. Nosotti, A. Zanella, P. Rebulla, *Banking together. A unified model of informed consent for biobanking*, in *EMBO Reports*, 9, 4, 2008 April, 307-313.

development of prevention in this area is undoubtedly a noteworthy factor in the development of the entire health and social system of the country. The greater the relevance of the screening programme to the overall system, the more urgent will become the management of the problematic aspects concerning the coordination of screening programmes at international and European level, both as regards the clinical usefulness of the tests and, consequently, the panel of the diseases to be checked and with regard to the management of the procedures for accessing the investigations. In addition, the issue of a general harmonisation of the Italian rules of 2016 must be urgently addressed in order to overcome the critical issues highlighted in the discussion and to respect the Italian constitutional principles ruling the health sector. In particular, in the Italian context, in the face of research and medical science that have developed scientifically valid screening programmes, we have traced some problems relating to the management of informed consent and dissent to the procedure deriving from some inconsistencies existing in the rules established in 2016 at national level. These inconsistencies have generated evident confusion at regional level about the voluntary or mandatory nature of the proposed screening tests with repercussions for the management of informed consent and dissent.

In this paper we have tried to outline a proposal for a constitutionally oriented reading of the rules of 2016 starting from the constitutional principles and the constitutional case law in the field of health protection.

As we observed, the constitutional principles enshrined in the Italian Constitution and the constitutional case law are clear to declare that only a State law, i.e. a primary source, can declare a treatment or a test voluntary, even by not specifying anything on the point of obligation and therefore enhancing the constitutional principle of self-determination, or mandatory, when there is the need for balancing individual right to health and collective interests to health.

In the case of newborns, there is a need for balance between the protection of the best interests of the child, the exercise of parental responsibility, and the collective interest in preventing serious diseases whose management strongly impacts, from a socio-economic viewpoint, on the community.

Collectively, the Italian rules of 2016 have essentially made it possible to move from a hybrid model – in which, with respect to three diseases at national level, on the basis of law n. 104 of 1992, the mandatory nature of tests was envisaged, while at regional level the broader diagnostic offer was based on the voluntary nature of the investigations – to an opting out model allowing for the implementation of uniformity of practice regarding tests and procedures.

The uniformity achieved in terms of the diagnostic offer through the rules of 2016 is still lacking regarding the awareness of the effects of the mandatory test and the organizational management of informed consent and dissent, and in terms of the collection of informed consent for the aspects not envisaged by the legislation of 2016.

Finally, on this latter aspect with regard to the issues of conservation of the biological material collected and its use for research purposes, there is a need to develop further regulatory guidelines that will make it possible to standardise regional practices on the subject and to guarantee the widest protection of fundamental rights of newborns and their families.

Special issue

In ordinary times, in extraordinary times: Consent, newborn screening, genetics and pandemics

Roger Brownsword, Jeffrey Wale*

ABSTRACT: Against the backdrop of newborn genetic screening and pandemics, this article examines disputes between parents, acting as proxies for their children, and healthcare professionals. While some will support parents, others will push-back against proxy consent and the right to veto actions that are proposed by the professionals. Whereas in ordinary times, such a push-back might seek to displace or downgrade parental rights (e.g. by appealing to professional duty or the optimisation of health) or to de-centre or dilute consent, in extraordinary times, rights and consent are superseded by appeals to responsibility, solidarity, and even "states of exception".

KEYWORDS: Proxy consent; newborn screening; genetic screening; stewardship; extraordinary times

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1. Introduction

Governance of the relationship between parents, acting on behalf of their children, and healthcare professionals, can be challenging; and, as is well-known, particular disputes can be emotionally charged and protracted.[1] While some will support the rights of parents, others will push-back against parents having the right to veto actions that are judged by the professionals to be either in the interests of the child or in the general interest. Whereas, in what we will call *ordinary* times, those who challenge parental rights and consent might appeal to, say, professional duty and to the optimisation of the health and well-being of the child, in *extraordinary* times, these familiar appeals are superseded by appeals to responsibility and solidarity, to unprecedented circumstances, and even to the dangerous idea of "states of exception".[2]

With a view to clarifying the nature of such contestation around parental rights and consent, our discussion is in three principal parts. First, we sketch our thinking about consent in general and proxy consent in particular. At the heart of this thinking is an "ideal-typical" model of proxy consent, set in a rights-focused regulatory context. This model provides a benchmark not only for mapping where particular regimes of medical law and public health law stand (both on paper and in practice) but also for highlighting four key ways in which pressure might be applied to the model: namely, with a view to *displacing* the rights-based paradigm within which consent plays its distinctive role, to *downgrading* the importance attached to, or the scope of, a particular right, to *de-centring* consent as a necessary and sufficient condition for justified action, or to *diluting* the particular requirements for a valid consent.

Secondly, assuming ordinary times and the typical pattern of contestation in biolaw and bioethics, we focus on newborn screening (NBS) programmes, some of which treat screening as mandatory (displacing both rights and consent), some of which operate with opt-out consent (diluting the signalling requirements for consent), and some of which (in line with our model) require a parental consent by opt-in. In the context of proposals to extend NBS so that more genetic data is obtained,[3] we seek to clarify the challenges to regimes of rights and, concomitantly, to our model of consent – not only the obvious challenges presented by the ethical approaches (whether utilitarian, paternalistic, or communitarian) that underlie regimes that are mandatory but also the challenges for

[1] Famously, see the case of Charlotte Wyatt and the Nuffield Council on Bioethics, *Critical care decisions in fetal and neonatal medicine: ethical issues* (London, November 2006). Recent examples are the cases of Charlie Gard and Alfie Evans, on the former of which see, e.g., E. CAVE, E. NOTTINGHAM, *Who Knows Best (Interests)? The Case of Charlie Gard, in Medical Law Review,* 26, 2018, 500; and J. MONTGOMERY, *The "tragedy" of Charlie Gard: a case study for regulation of innovation*, in *Law, Innovation and Technology,* 11, 2019, 155.

[2] On which, see A. SUPIOT, *The Return of "Rule by Men",* in ID., *Governance by Numbers*, Oxford, 2017, (trans Saskia Brown).

[3] For assessment, see, e.g, B.A. TARINI, A.J. GOLDENBERG, *Ethical Issues with Newborn Screening in the Genomics Era,* in *Annual Review of Genomics and Human Genetics*, 13, 2012, 381; and, S. TAYLOR-PHILLIPS et al., *The Ethical, Social and Legal Issues with Expanding the Newborn Blood Spot Test,* Warwick, 2014, (on file with authors). On the potential of extending NBS, see, UK National Screening Committee, *Generation genome and the opportunities for screening programmes*, 2019, 18-19, available at: https://bit.ly/3aLDztR (last accessed October 11, 2020); and HM Government, *Genome UK: The future of healthcare*, September 2020, 27-28, available at: https://www.gov.uk/government/publications/genome-uk-the-future-of-healthcare (last accessed October 11, 2020).

ISSN 2284-4503

rights theorists of doing justice to the model of consent as well as ensuring that parental rights are not abused.

Thirdly, assuming extraordinary times, we consider disputes between parents and healthcare professionals with particular reference to public health interventions in the context of a pandemic (for example, concerning the vaccination of children or their participation in a research study to identify relevant genetic markers). Resisting the framing of such disputes in a way that treats extraordinary times as a state of exception (giving professionals a carte blanche), or as an extension of ordinary times arguments (but with utilitarian and duty-based arguments for solidarity now having the upper hand as against rights-based arguments and individual consent), we argue for a quite different understanding of the issues raised by extraordinary times considerations. Crucially, we argue that our best understanding of concerns about responsibility and solidarity is that ordinary time justifications are superseded in extraordinary times by the urgent need to take steps to restore and maintain the global commons (the pre-conditions for the existence of any kind of human community with any kind of guiding ethical perspective). Accordingly, even for a community that, in ordinary times, takes both rights and consent seriously, in extraordinary times, it will be the stewardship responsibilities relating to the global commons that supply the overriding reasons for action.

Finally, in some short concluding remarks, we indicate some of the further questions that are invited by, and the implications to be drawn from, our analysis.

2. Consent and Proxy Consent

Before sketching an ideal-typical model of proxy consent, that model itself needs to be placed within the context of a distinctively rights-based regime of rules and standards. Accordingly, we start with some prefatory remarks about such a regime and the justificatory function of consent within it before setting out our ideal-typical model of proxy consent and the principal pathways to deviation from it.[4]

2.1. The paradigm of a rights-based rule regime

The paradigmatic setting for consent is a rights-based regime of rules and standards (legal or moral) that regulates the interactions and transactions between individual agents as well as the relationship between citizens and public bodies. In such a regime, individuals have claim rights (that some other individual or individuals should or should not do x); and, it follows that those latter individuals have a correlative duty to do, or not to do, x.

The function of consent is both dynamic and justificatory. Consent is dynamic in the sense that it enables rights-holders to alter their position in relation to duty-bearers by waiving the *benefit* of a right although not the *right* itself; and, consent is justificatory in the sense that it authorises an act that would otherwise violate a right. However, consent does not suffice to show that an action is right as such; rather, it suffices to prevent a complaint (by the consenting party) that an action involves a violation of the consenting party's rights. Hence, where A consents to some action, x, by B,

[4] In this part of the article, we draw on D. BEYLEVELD, R. BROWNSWORD, *Consent in the Law,* Oxford, 2007.

then the function of that consent is to authorise B to do x and, at the same time, to preclude a complaint by A that B thereby violates A's right that x not be done. However, A is the only party so precluded – or, at any rate, this is the case unless A is acting as an agent or as a proxy for others. So, for example, if B's doing x is a prima-facie wrong relative to Z, the fact that A has consented to B doing x is no answer to Z. Where A is acting as a proxy, a similar analysis applies. Thus, where A, in consenting to some action, x, by B is doing so as a proxy for C, we take it that A's consent precludes a complaint by both A and C that B has done wrong by doing x. In both cases, consent serves, as Lord Donaldson famously expressed it, as a flak-jacket for B, but the cover that it provides for B is limited to the consenting parties.[5]

Turning this round, if B goes ahead and does x (which is covered by A's right) without having obtained A's consent (indeed, where A has explicitly declined to give consent), then B seems to have no answer to A's complaint that B has infringed A's right. So far, so straightforward. However, in a rights-based regime, B might concede that doing x does involve a violation of A's right (which, moreover, is a matter for regret) but that the doing of x is justified all things considered because it serves a higher ranking right than the right violated.

This formal analysis leaves much still to be settled. For example, there are important questions about the rights-based regime itself, about the substance and scope of particular rights, about how conflicting rights are ranked and prioritised, and so on; and it might fairly be said that, if parental rights are to be privileged and if their consent is to be protected by stringent requirements, we really need to understand why such importance is attached to these particular interests. These are matters to which we will return in the next part of the article.

2.2. An ideal-typical model of consent by proxy

In principle, there are many models of consent – broad and narrow, opt-in or opt-out, static and dynamic, relational and non-relational, and so on.[6] For the purposes of anchoring our discussion, and providing a benchmark for various regimes of proxy consent, we will sketch an ideal-typical model of consent by parental proxy – that is, a model where parents have a veto and where their consent is taken seriously as the justifying reason for whatever action, x, is proposed in relation to their baby (such as drawing, analysing and storing a baby's blood).[7]

Bearing in mind that our model is placed in the setting of a rights-based regime of rules, and assuming that there are no doubts about the capacity of the parties, the central features or conditions of the ideal-type are as follows:

[5] See, *Re W (A Minor) (Medical Treatment: Court's Jurisdiction)* [1993] Fam 64.

[6] See, e.g., R. Brownsword, *Regulating Biobanks: Another Triple Bottom Line*, in G. Pascuzzi, U. Izzo, M. Macilotti (eds), *Comparative Issues in the Governance of Research Biobanks,* Heidelberg, 2013, 41; J. Kaye, E.A. Whitley, D. Lund, M. Morrison, H. Teare, K. Melham, *Dynamic consent: a patient interface for twenty-first century research networks*, in *European Journal of Human Genetics,* 23, 2015, 141; and I. Budin-Ljøsne et al., *Dynamic Consent: a potential solution to some of the challenges of modern biomedical research*, *BMC Medical Ethics,* 18, 2017, available at: https://bit.ly/2RNFwPv, doi: 10.1186/s12910-016-0162-9.

[7] Nb, although we are presenting this model as an "ideal-type", we are not claiming that this approach to consent is "ideal", merely that it is a robust representation of the view that the consent of the parents really matters.

- Consent by the parents is viewed as the necessary and sufficient justifying condition for the proposed action, x; and, as a corollary, where parents either decline to give their consent, or simply do not give their consent, then doing x will involve a violation of the parents' prima facie rights.
- The parents' consent is to be treated as valid only where it is (a) explicit and clearly signalled, (b) freely given and (c) informed; and,
- The consent of the parents should, at least as a matter of aspiration, flow from a relational process.

The significance of the first condition – the sovereignty of consent – is that those who take action x have one, and only one justifying answer to the parents (and to the baby or child on whose behalf the parents act as proxy) where it is claimed (and conceded) that action x involves a violation of the parties' rights. That answer is that the parents consented to x. It is no answer, for example, to appeal to professional duty or social utility. Of course, as we have already flagged up, within the rights-based paradigm, the violation of the parties' rights might be admitted but an all things considered justification might be put forward. So, we need to be careful about how we express the sovereignty of consent. In a rights-based regime, consent is the necessary and sufficient justification for an act that involves a prima facie violation of a right but that (wrongful) act might still be justified by appealing to a more compelling right. The significance of the second condition only becomes apparent when we spell out the demanding nature of the specification of a valid consent. In other words, it is only when we specify that the signalling of consent must be unequivocal, that the circumstances in which consent is given must be free of undue or improper pressure, and that being informed is not satisfied merely by the absence of misinformation, that we grasp that the ideal-typical model permits no short cuts to a valid consent. As for the third condition, although this is aspirational and does recognise some practical limitations, it clearly views consent as more than a singular event. No doubt, the term "consent" is often used to denote a specific transaction between stakeholders or to fix the particular moment when A authorises B to do x (thereby exercising a power to transform B's duty not to do x into an immunity where x is done).[8] However, in practice, to the extent that this mode of thinking tends to encourage a perfunctory approach to consent, it needs to be adopted with care.

Finally, two other points, should be noted. One point is that it is one thing for a person to give or to withhold consent purely on their own behalf and quite another thing for parents acting as a proxy for their baby or child to give or withhold their consent. In the former case, it is simply the interests of the person that are relevant – and, indeed, we might say that their decision to give or to withhold consent, no matter how irrational or unreasonable it might seem, is to be respected.[9] By contrast, if a parental decision to give or to withhold their proxy consent seems to be irrational or unreasonable relative to the (clearly relevant) interests of the baby or child, then we are likely to take a very

[8] Compare W.N. HOHFELD, *Fundamental Legal Conceptions*, New Haven, 1964.

[9] As Lord Donaldson MR put it in *Re T (adult: refusal of medical treatment)* [1992] 4 All ER 649, 663: "the patient's right of choice exists whether the reasons for making that choice are rational, irrational, unknown or even non-existent. That his choice is contrary to what is to be expected of the vast majority of adults is only relevant if there are other reasons for doubting his capacity to decide".

different view of whether this is within the scope of the parental rights or whether it should be respected. The question of how the interests of the child might constrain parental rights is a matter to which we will return in section 3.4. The other point is that the distinction between persons consenting on their own behalf and parents consenting as proxies might be relevant in the event of there being some doubt about how to interpret the scope and breadth of the consent that is given. Arguably, in cases of doubt, the default should be a restrictive approach; but, where we are dealing with parents consenting as proxies, a contextual reading might be more appropriate. In practice, this point of interpretation might be an important one and it merits further consideration than we can give it here.

2.3. Four deviations from the ideal-typical model

Where there is pressure on parents to give their consent to action x, there is more than one pathway to deviation from the ideal-type. The four principal pathways are: displacement, downgrading, de-centring, and dilution.

First, pressure might be applied directly or indirectly (via consent) to *displace* the rights paradigm itself. Instead of rights, it might be argued, for example, that actions should be justified by reference to the duties and responsibilities that individuals have (to themselves and to others), or by some desirable consequences such as the maximisation of utility, or the minimisation of distress, or by the reduction of inequality, and so on.

Secondly, pressure might be applied directly or indirectly (via consent) with a view to *downgrading* the scope or significance of a particular right. In principle, this is quite different to arguing for the displacement of the rights paradigm; but, in practice, the flattening or narrowing of a particular right might be part of a more general ambition to challenge the rights paradigm.

Thirdly, in relation to the first condition of our model, pressure might be applied with a view to *de-centring* consent. Here, the argument would be that consent by the parents might be treated as sufficient but not strictly necessary – in just the way, for example, that, in some data protection regimes, the consent of data subjects to the processing of their personal data is a sufficient but not a necessary condition for lawful processing.[10] It follows that, even if the parents have not given their consent to the doing of x, the doing of x might not be viewed as any kind of wrong. Consent becomes simply one justifying option; and, the withholding of consent is no longer a serious blockage.

Fourthly, pressure might be applied to the second condition of our model with a view to *diluting* the requirements for a valid consent (or, possibly, as a strategy to displace or downgrade rights in practice). For example, there might be pressure to amend the specification so that implicit and indistinct signalling will suffice (as in opt-out models of consent); or it might be the requirement of freely given consent that is diluted; or the pressure point might be the informational requirement.

Of these four deviations, it is displacement that goes for the jugular. Sometimes, the seriousness of the challenge might be masked because the point of attack seems to be on consent itself; but, on closer analysis, we see that it is the rights-based regime that is actually the target.[11] Conversely, we

[10] Cf. the UK Data Protection Act, 2018.

[11] Compare, R. BROWNSWORD, *Rights, Responsibility and Stewardship: Beyond Consent,* in H. WIDDOWS, C. MULLEN (eds), *The Governance of Genetic Information: Who Decides?*, Cambridge, 2009, 99.

might think that dilution is the least threatening challenge. However, in practice, relaxation of the conditions for a valid consent can mean that those who seek consent are pushing at an open door. What we end up with then is a regime that pays lip service to both rights and consent but which actually has a quite different orientation. Once the specification of a valid consent is weakened in these ways, we might find that consent, far from protecting the interests of the intended party, becomes a convenient way for medical professionals or healthcare researchers to "legitimate" their actions. Ultimately, advocates of a proxy consent framework need to be alert to the full spectrum of challenges, both to the underlying rights paradigm and to the particulars of consent.[12]

3. In Ordinary Times: Proxy Consent and Newborn Genetic Screening

In this part of the article we discuss ordinary time debates about the extension of NBS programmes (in order to capture more genetic data). Our particular interest is both in mapping where regimes stand relative to our model of consent and understanding how regimes that are publicly committed to consent can be put under pressure to defect from those commitments.

Our discussion proceeds in four stages as follows: (i) we sketch the typical pattern of debates and disputes in modern biolaw and bioethics, where rights-based approaches are confronted by a plurality of rival approaches each of which, in its own way, challenges the importance and significance attached to consent; (ii) we distinguish between three models of NBS programmes – mandatory, opt-out, and opt-in, the latter two of which purport to rest, to a greater or lesser degree, on the sovereignty of consent; (iii) we consider the range and potential complexity of decisions to be made by the parents where NBS extends the genetic data collected; and (iv) we consider the legitimate scope of the rights of parents in making decisions as proxies for their children.

3.1. The pattern of debates in biolaw and bioethics

It was more than 50 years ago that Wilson and Jungner identified ten core principles for screening programmes.[13] Three aspects of their seminal statement are striking. First, the principles set a high bar for investment in screening – not only should the conditions screened for be important and treatable, screening for those conditions needs to be effective and acceptable. Secondly, and not surprisingly, the principles do not speak to genetic screening. Thirdly, perhaps a little more surprisingly, the principles do not say that screening programmes should be based on informed consent, let alone the proxy consent of parents where children are screened[14]. While these principles

[12] Compare, ID., *Consent in Data Protection Law: Privacy, Fair Processing, and Confidentiality,* in S. GUTWIRTH, Y. POULLET, P. DE HERT, C. DE TERWANGNE, S. NOUWT (eds), *Reinventing Data Protection?*, Dordrecht, 2009, 83.

[13] J.M.G. WILSON, G. JUNGNER, *Principles and practice of screening for disease,* Geneva, 1968.

[14] That said, it might be argued that (i) because we are discussing population-wide screening programmes rather than individual testing, we are already in the realm of public health, from which it follows (ii) that this is a matter for judgments about the public good rather than private right (and consent). However, quite apart from any difficulty in differentiating between screening and testing, the second part of the argument clearly begs the question. The fact that decisions are being made for the public good or in the public interest does not mean that individual rights are no longer relevant. See further 3.4.2.

have been refined and added to,[15] the culture of screening is still relatively cautious which suggests that advocates of expanded NBS, before engaging with the larger community, would first have to persuade the screening community that the proposal makes sense. How might the arguments go?

The pattern of debates in ordinary time biolaw and bioethics often has the following shape. The general direction of travel is towards recognising the centrality of individual rights (rights of patients and research participants) and the importance of informed consent. However, a rights-based ethic (with our ideal-typical model of consent) finds itself challenged by utilitarians (who regard consent as a transaction cost to be minimised in the pursuit of general benefit), paternalists (who believe that they should be the judges of the best interests of others) and conservative dignitarians (who deny the relevance of both rights and consent). While there might be some convergence between these approaches, there is a standing tension between them.[16] It also should be said that, within each ethical "camp", there will often be significant differences (both as to the relevant principles and as to their application to particular cases). For example, amongst utilitarians there might be different assessments of the likely benefits and harms resulting from an extension of NBS; and, while some dignitarians will be communitarians, others will be Kantians, and yet others will be relying on the doctrinal teaching of the Catholic Church.

This thumbnail sketch does not suffice to predict exactly how debates about the extension of NBS will go in any particular community. However, we might expect a degree of convergence between the positions taken by utilitarian and rights-based ethicists – at any rate, to the extent that there is a shared interest in making more actionable information available to parents. For utilitarians, this is a net benefit; and, for rights-theorists, it is in line with recognising parental autonomy (possibly articulated in terms of a right to know). Nevertheless, the convergence is far from complete. For those who take consent seriously, the information must be at the option of the parents; it is their choice whether or not to sign up for the extended NBS. So, when the extension of NBS is proposed, rights-based ethicists might give it their qualified support, their emphatic caveat being that parents should continue to be in a position to say yes or no on a free and informed basis.

By contrast, some paternalists might argue that it is not in the best interests of either parents or their children to have this kind of information; and, while some dignitarians might support a mandatory screening programme (as a matter of solidarity within the community), they might oppose the further "geneticization" of society on the grounds that it compromises human dignity. Given this pattern, it will be paternalists and dignitarians who seek to displace or downgrade parental rights and it will be utilitarians who, to the extent that they are persuaded that an extension of NBS will be beneficial, focus primarily on the displacement or dilution of parental consent.

[15] A. ANDERMANN, I. BLANCQUAERT, S. BEAUCHAMP, V. DÉRY, *Revisiting Wilson and Jungner in the genomic age: a review of screening criteria over the past 40 years,* available at: https://www.who.int/bulletin/volumes/86/4/07-050112/en/ (last accessed April 29, 2020).

[16] Compare R. BROWNSWORD, *Bioethics Today, Bioethics Tomorrow: Stem Cell Research and the "Dignitarian Alliance",* in *University of Notre Dame Journal of Law, Ethics and Public Policy* , 17, 2003, 15; and ID., *Rights, Regulation and the Technological Revolution,* Oxford, 2008.

3.2. Contemporary models

NBS programmes might be placed in one of three categories: (i) where screening is mandatory; (ii) where screening is "normal" (or "advised") but subject to opt-out; and (iii) where screening is by opt-in.

In principle, the distinction between regimes that require consent and those that do not should be clear, as should the distinction between consent-based regimes that require opt-in and those that treat opt-out as sufficient. However, in practice, the lines between these different regimes can become blurred. Not only does this make categorisation more difficult, it can obscure the significance of the headline distinctions. Whereas, in principle, the distinction between mandatory regimes (where neither rights nor consent are taken seriously) and non-mandatory regimes (where parental refusal to NBS is determinative) is of capital importance, the difference between *opt-in* and *opt-out* models might seem to be a matter of detail. However, opt-out, rather than opt-in, might already reflect some dilution of the requirements for a valid consent and, indeed, the practical reality might be that, de facto, NBS is as good as mandatory.

Bearing in mind these cautionary remarks, we can speak briefly to each of the three models before conducting an initial stock-taking of how this bears on a proposed extension of NBS programmes.

3.2.1. Mandatory models

On the face of it, by requiring NBS, mandatory models displace parental rights and their proxy consent.[17] As FIGO (the International Federation of Gynecology and Obstetrics, and the self-proclaimed global voice for women's health) has put it: "in view of the fact that the overall acceptability of NBS is beyond doubt, NBS should be mandatory and free of charge if early diagnosis and treatment will benefit the newborn".[18]

Precisely which ethic supports this position is open to interpretation: as stated, it is in line with utilitarian thinking but it could be read as a paternalistic or even a communitarian stance that infers that parents do not have a legitimate or sufficient priority interest in the screening process. In many cases, though, this position is qualified by a State obligation to make relevant information available to parents, and by the need for separate written consent for other uses (including retention for research).[19] Not only does this highlight a distinction between obtaining up front agreement to testing/reporting, and agreement to the subsequent retention and use of samples/data, it foreshadows the potential complexity of mapping NBS regimes by reference to the role of consent.

[17] However, in some mandatory regimes (such as those operating in some US States) there will be a "conscientious objection" clause that permits parents to opt-out. See, B.A. TARINI, A.J. GOLDENBERG, *op.cit.*; and, J.R. BOTKIN, *Ethical Issues in Newborn Screening*, in L. FRANCIS (ed), *The Oxford Handbook of Reproductive Ethics*, Oxford, 2017, 251.

[18] Figo Committee Report, *Ethical aspects concerning neonatal screening FIGO Committee for the Ethical Aspects of Human Reproduction and Women's Health*, in *International Journal of Gynecology and Obstetrics*, 106, 2009, 273-274. However, while it might be "beyond doubt" that NBS is acceptable in the established cases, it is surely an over-statement if whole genome or exome screening is to be added.

[19] *Ibid*.

3.2.2. Opt-out models

In opt-out models, parents are nudged towards NBS. State actors often endorse or promote NBS, with parents retaining an option or choice not to test. Although there is implicit recognition that the parents have a legitimate interest in the process, screening is the norm. Parents bear the primary burden because they need to take active steps to exercise the option not to screen. The degree of burden may vary and opt-outs may be especially demanding if complex bureaucratic and explanatory mechanisms are put in place. Moreover, these already sticky nudges might be compounded by over-enthusiastic promotion by health care professionals, "hectic" environments,[20] tired parents, variability in comprehension and discretionary action by healthcare professionals, all making these models vulnerable to failure, especially in expanded frameworks with multiple conditions and options. Accordingly, while an opt-out model might purport to take parental rights and proxy consent seriously, in practice, it is all too easy to see how such a regime might both de-centre and dilute consent.

3.2.3. Opt-in models

Where opt-in models are adopted, nothing short of explicit, clearly signalled, free and informed proxy consent that covers all stages – including testing, the return of findings, subsequent use and storage – should suffice. All conditions in our ideal-typical model need to be met if we are serious about demonstrating that parents not only have a legitimate, but also an essential interest in screening. That said, in cases of parental conflict or indecision, there might be some nice questions about whether both parents must consent or agree to withhold consent. There may also be cases where parental consent is not available or where the State judges that there are "good" (compelling rights') reasons to override the parental position.

England promotes an "opt-in" model for NBS using a verbal only and single parent proxy consent process, supported by an informational leaflet and initial discussion in the third trimester of pregnancy.[21] Bloodspot screening is carried out for up to 9 conditions, with some parental choice over the testing and report options.[22] Parental proxy consent is formally recorded in the newborn records, and additional written communication is required when parents decline testing (including notification to the GP and Child Health Information Service). The Public Health England (PHE) letter template – intended for parents declining blood spot testing – says that, while "screening is not compulsory, […] it is strongly recommended because it could save your baby's life".[23] This is then re-inforced by the warning that, unless parents change their minds about NBS, "there is a risk that your

[20] R. Botkin, *Ethical Issues in Newborn Screening*, cit., 255.

[21] Public Health England (PHE) issues many publications about newborn screening, many of which are aimed at healthcare professionals, and some of which are highly technical: see https://bit.ly/2HyWAnV (last accessed August 14, 2020). Information specifically for parents can be found at https://bit.ly/3awILBo (last accessed August 14, 2020).

[22] F. Ulph, S. Wright, N. Dharni, et al., *Provision of Information about newborn screening antenatally: a sequential exploratory mixed-methods project,* in *Health Technol,* 21, 55, 2017, Assess 1. Parents can only choose to have screening for all 6 Inherited Metabolic Diseases or none at all (PHE n 21).

[23] Available at, https://www.gov.uk/government/publications/declined-newborn-blood-spot-screening-template-letters (last accessed August 14, 2020).

child may become seriously ill and suffer irreversible harm".[24] The reasonable inference is that responsible parents would and should consider testing. Evidence shows that fathers are not always involved in the consent process[25] and, as we have already remarked, we cannot rule out the possibility of family discord.[26] Some concern has been expressed about the routinisation of testing and the inadequate provision of information to parents.[27] This suggests that some dilution of consent may be occurring in practice, either because the signalling is cursory or because the parents do not have adequate information to make an informed choice. Some aspects of the process – for example, whether to be involved in future research – appear to require a positive opt-out by the parent, suggesting that differential standards and interests are in play.[28]

Given this background, we might wonder whether the English model is actually closer to the *opt-out* variant. Two recent studies of the English NBS programme[29] shed some light on the matter. In the first of their studies, Ulph et al. note that the timing of information provision is critical to its effectiveness – with informational exchange shortly after birth being generally ineffective and third trimester exchange being a key time for informational assimilation. This study suggests that NBS design should focus on the informational needs of parents rather than upon the process of obtaining and formalising consent. In their second, qualitative, study Ulph et al. sought the views of healthcare professionals and parents about NBS consent processes in England. There was common consensus that dilution of consent was occurring in practice (this being expressed as doubts about the voluntary and informed aspects of the process). There was also some evidence that parents valued disclosure of information more than choice – with parents being happy to have any tests "for the health of their baby".[30] However, future retention and research use generated specific concerns about trust and differential views about consent,[31] suggesting that unitary approaches to consent may not be what parents want in practice.

3.2.4. Taking stock: extending NBS

How might a proposed extension of NBS (to collect genetic information) sit with the contemporary models that we have identified? Three points seem to be noteworthy.

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[24] *Ibid*.

[25] F. Ulph, N. Dharni, R. Bennett, T. Lavender, *Consent for newborn screening: screening professionals' and parents' views*, in *Public Health,* 178, 2020, 151, 156.

[26] C.A. Genetti, T.S. Schwartz, J.O. Robinson, *Parental Interest in Genomic Sequencing of Newborns: Enrollment Experience from the BabySeq Project*, in *Genetics in Medicine,* 21, 3, 2019, 622–630, doi:10.1038/s41436-018-0105-6, 6.

[27] F. Ulph, S. Wright, N. Dharni, et al., *Provision of Information about newborn screening antenatally: a sequential exploratory mixed-methods project*, cit.; F. Ulph, N. Dharni, R. Bennett, T. Lavender, *Consent for newborn screening: screening professionals' and parents' views,* cit.

[28] See https://bit.ly/2HyWAnV.

[29] F. Ulph, S. Wright, N. Dharni, et al., *Provision of Information about newborn screening antenatally: a sequential exploratory mixed-methods project*, cit.; F. Ulph, N. Dharni, R. Bennett, T. Lavender, *Consent for newborn screening: screening professionals' and parents' views,* cit.

[30] F. Ulph, N. Dharni, R. Bennett, T. Lavender, *Consent for newborn screening: screening professionals' and parents' views*, 154.

[31] *Ibid*.

First, it might be thought that where NBS is mandatory, there will be little resistance. However, the proposed extension will need to be squared with whatever background justification for screening is operative in the community. Even where the screening community is guided by whether there is a net benefit, there might be reservations about extending NBS in the absence of clear benefit to the child.

Secondly, if the benefits of an extension to NBS are uncertain, this has implications not only for benefit-focused mandatory regimes but also for advocates of parental rights and consent.[32] As the benefits to the test subject dwindle and any correlative parental duty to screen is weakened, the ability to deliver workable informed consent processes becomes more problematic. The expression of gene variation is shaped by a range of internal/external factors that make whole exome or genome results difficult to interpret or to counsel upon either in advance or once results are available.[33] Moreover, although parents might be given more screening options to which they can say yes or no, the complexity of the choices presented may mean that, while parents are able to make more decisions, the decisions that they make are less meaningful.[34] In short, more choice coupled with more information does not necessarily translate into better choices.

Thirdly, creating what are in effect genetic profiles for the newborn, raises familiar broader concerns about the social impact of genetic information.[35] Whatever our ethics, these broader concerns apply. Capturing these concerns, a Hastings Center Report says that "mapping and classifying people's genomes would undermine their privacy; lead to new forms of discrimination (by employers and insurers, for example); foster the essentialist idea that people are their genes; bolster attempts to interpret social identities in biological terms; and trigger depression, anxiety, suicidality, and worry in individuals whose genetic risk for certain conditions was determined to be high. In addition, some cautioned that a genetics focus would lead to lessened emphasis on the social determinants of health and health disparities among underserved groups (such as racial or ethnic minorities)".[36]

In sum, we should not assume either that those who advocate the extension of NBS will be pushing at an open door in mandatory regimes, or that those who are trying to operationalise an opt-in model (along the lines of the ideal-type) will find it easy to do so. Possibly, the most likely defection from the ideal-typical model will be in those regimes that pay lip service to opt-in but which, in practice, are actually closer to opt-out. In this context of parents being already nudged towards consent, practice is likely to fall short of the aspirational relational process, and the information given to parents is likely to be unsatisfactory.

[32] See, e.g., .R. BOTKIN, *Ethical Issues in Newborn Screening,* cit., 260.

[33] J. JOHNSTON et al., *Sequencing Newborns: A Call for Nuanced Use of Genomic Technologies, The Ethics of Sequencing Newborns: Recommendations and Reflections, special report,* in *Hastings Center Report,* 48, S2, 2018, DOI: 10.1002/hast.874. See also K.H. ROTHENBERG, L.W. BUSH, *The Drama of DNA: Narrative Genomics*, Oxford, 2014.

[34] G. DWORKIN, *The Theory and Practice of Autonomy*, Cambridge, 1997; F. ULPH, S. WRIGHT, N. DHARNI, et al., *Provision of Information about newborn screening antenatally: a sequential exploratory mixed-methods project,* cit.

[35] See, e.g., Human Genetics Commission, *Profiling the Newborn: a Prospective Gene Technology*?, January 2005.

[36] J. JOHNSTON et al., *op.cit.,* S16.

3.3. The range of decisions to be made around NBS

For the screening community and policymakers, it is not just a matter of deciding whether or not to offer NBS; and, concomitantly, for parents it is not just a matter of saying yes or no to NBS. There are decisions to be made about which particular diseases and conditions to screen for, about the return of findings, and about the retention of data and samples.

First, a decision has to be made about whether to limit NBS to treatable/remediable conditions, or extend it to a range of late onset conditions or carrier conditions. In part, this may be influenced by the analytical validity,[37] clinical validity[38] and the clinical utility[39] of the screening test. The limitation of many NBS programmes to treatable conditions provides an obvious fulcrum for activism,[40] but keeps the focus firmly on the direct interests of the test subject and makes the benefit/harm assessment much easier.

Second, a decision has to be made about which findings are reported back by the laboratories and retained in the data records. Laboratories may control the reporting/recording process, although this will probably be determined or influenced by health policies or instructions from the healthcare professionals and/or parents. Decisions have to be made about whether to report back incidental findings – where information about a condition or risk is identified but was not the subject or original purpose of the test.[41] This gives rise to a vexed discussion about the existence of a right to know and right not to know, and to the respective rights' holder in this context.[42] Decisions have to be made about what happens to the retained and unfiltered findings.[43] There might be concerns about having any findings, uncertain or otherwise, permanently linked to a child's medical or other data records and what implications it might have for their future.[44] Further, there is a question about whose interests should feature and be prioritised at this stage of the process. Even if newborn interests have primacy at the "decision to test" stage, the interests of other family members might need to be brought into account once there are incidental findings that concern and potentially benefit them.[45] If reporting back to the family reduces the diagnostic odyssey, or enables parents to be forewarned

[37] The ability to detect the trait/condition it seeks (sensitivity and specificity).

[38] The predictive accuracy of the test.

[39] The ability and usefulness of any test to improve the health/wellbeing of the person tested.

[40] Compare, the Genetic Alliance UK, *Fixing the present, building for the future: Newborn screening for rare diseases*, 2019, 16, where it is claimed that the UK screens for relatively few diseases compared to other high-income countries.

[41] Generally, see S. VAN DER BURG, A. OERLEMANS, *Fostering caring relationships: Suggestions to rethink liberal perspectives on the ethics of newborn screening,* in *Bioethics,* 32, 2018, 171.

[42] For discussion: J. WALE, *Regulating disruptive technology and informational interests in the arena of reproductive tests,* in *Journal of Information Rights, Policy and Practice*, 3, 1, 2019, available at: https://jirpp.winchesteruniversitypress.org/articles/abstract/24/; R. BROWNSWORD, J. Wale, *The Right to Know and the Right Not to Know Revisited*, in *Asian Bioethics Review*, 2017, 1-16, doi:10.1007/s41649-017-0012-1; B DAVIES, *The right not to know and the obligation to know*, *Journal of Medical Ethics,* 2020, 1-4, doi:10.1136/medethics-2019-106009.

[43] B.A. TARINI, A.J. GOLDENBERG, *op.cit.*; Genetic Alliance UK, *Fixing the present, building for the future: Newborn screening for rare diseases*, cit., 18.

[44] C.A. GENETTI, T.S. SCHWARTZ, J.O. ROBINSON, et al., *op.cit.*

[45] J. JOHNSTON et al., *op.cit.*

of possible risks in future pregnancies,[46] those who espouse a utilitarian or communitarian ethic might be attracted by a broad obligation to return incidental findings.[47] However, for those who aspire to keep faith with the ideal-typical model of consent, there is no easy option: whether the parents are invited to make a blanket yes or no decision about the return of findings, or asked to make a more nuanced choice, this looks like a major challenge for informed consent.

Third, there are decisions to be made about what data and samples are retained, for how long and for what purposes. FIGO have suggested that later sample use – for retrospective testing or research purposes – requires written permission. This is a more parent-centric position when compared to their stance on testing,[48] and is supported by empirical evidence showing that many parents prefer to make informed choices in the context of blood spot storage and subsequent research use.[49] In terms of existing laboratory data – whether reported back or otherwise – there are issues of privacy, trust, and possible future prejudice or discrimination.[50] England uses a default blood spot retention period of "at least 5 years", with standard use covering quality improvement and "research to help improve the health of babies and their families in the UK".[51] The screening results are also recorded in various health information systems. Identity is supposed to be anonymised if the samples are used for research purposes, and the parents have to formally opt out of research if they have agreed to screening. This highlights some ambivalence in screening policy and practice: namely, is screening for the benefit of the test subject, or is it to collect/retain samples for research purposes and for wider societal benefit?[52] Some might argue that if we can achieve both aims, there will be no wrong done to the test subject. However, this win-win argument implies a utilitarian approach that either displaces rights and consent or downgrades parental rights.

Pulling together the strands of this discussion, if NBS is extended in order to capture more genetic information, the potential complexity of decision-making will challenge the commitment to both the sovereignty and the validity of consent.[53] Where consent is formalised before testing in a single

[46] .R. BOTKIN, *Ethical Issues in Newborn Screening*, cit.

[47] See for eg., M.S. GROSS, A.R. RUTH, S.A. RASMUSSEN, *Respect women, promote health and reduce stigma: ethical arguments for universal hepatitis C screening in pregnancy*, *Journal of Medical Ethics,* 2020, 1-4 DOI:10.1136/medethics-2019-105692.

[48] Figo Committee Report, *Ethical aspects concerning neonatal screening FIGO Committee for the Ethical Aspects of Human Reproduction and Women's Health,* cit.

[49] F. ULPH, S. WRIGHT, N. DHARNI, et al., *Provision of Information about newborn screening antenatally: a sequential exploratory mixed-methods project,* cit.

[50] See for e.g., C.A. GENETTI, T.S. SCHWARTZ, J.O. ROBINSON, et al., *op.cit.*

[51] See https://bit.ly/2HyWAnV. Interestingly, the destruction of residual blood spot cards is currently embargoed while the associated Code of Practice is being reviewed (available at: https://www.nhs.uk/conditions/pregnancy-and-baby/newborn-blood-spot-test/).

[52] E.W. ROTHWELL, R.A. ANDERSON, M.J. BURBANK et al., *Concerns of Newborn Blood Screening Advisory Committee Members Regarding Storage and Use of Residual Newborn Screening Blood Spots*, in *American Public Health Association*, 2011, DOI 10.2105/AJPH.2010.200485.

[53] For example, see WHO, *Medical Genetic Services in Developing Countries: The Ethical, Legal and Social Implications of Genetic Testing and Screening,* 58, 2006, para 4.6.2, for the view that valid informed consent (in the context of genetic testing) "requires a bilateral process involving a dialogue of questions and answers between the individual considering testing and the person obtaining informed consent (often a health care professional). This dialogue requires the person obtaining informed consent to gauge the appropriate level of language and technical detail suitable for the individual's understanding".

(agreement) transaction, with informational exchange occurring shortly before and after birth, there are already concerns about informed consent; and so, we can have no confidence that these processes will improve in the event of expansion. While those with a reformist agenda have suggested that we may do better to focus on the informational needs of parents rather than the moment of consent itself,[54] others have mooted differential consent arrangements where the direct benefit test cannot be met.[55] A further option might be to break down decision-making and consent processes into more manageable elements. While there could be resource and uptake issues if parents were approached about retention/research options at a later date, the advantage may be that critical decision-making will be occurring in less hectic and more informed contexts. Alternatively, stratified informed consent processes could be used where the informational exchange is dependent on what parents want to know.[56]

While there is much to be said about these ideas, which clearly merit further consideration, we cannot explore them any further here. For present purposes, suffice it to say that the practical challenge of keeping faith with parental rights and the sovereignty of consent should not be under-estimated.

3.4. Legitimate interests and the scope of parental rights

It is one thing to argue that parents have a legitimate interest in making their own choices; but it does not follow that parents should be able to choose just what they want.[57] In a community that takes rights and consent seriously, there will be decisions to be made about the limits to, and scope of, parental rights and about the resolution of competing and conflicting rights.

3.4.1. Limiting parent autonomy

Most communities accept that parental autonomy is not absolute,[58] that some limits have to be be placed on parental freedom vis a vis their children.[59] Criminal frameworks typically regulate both positive conduct and omissions that harm or have the risk of seriously harming children. Regulation

[54] F. Ulph, N. Dharni, R. Bennett, T. Lavender, *Consent for newborn screening: screening professionals' and parents' views* , cit.; S. Van der Burg, A. Oerlemans, *Fostering caring relationships: Suggestions to rethink liberal perspectives on the ethics of newborn screening,* cit.

[55] B.A. Tarini, A.J. Goldenberg, *op.cit.*; but, concern has been expressed about the practical implications of these approaches, see J.R. Botkin, *Waving Goodbye to Waivers of Consent*, in *The Hastings Center Report*, 45, 6, 2015, DOI: https://doi.org/10.1002/hast.520.

[56] E.M. Bunnik, A. de Jong, N. Nijsingh, *The new genetics and informed consent: Differentiating choice to preserve autonomy*, in *Bioethics,* 27, 2013, 348-355.

[57] For discussion in the context of non-invasive pre-natal testing, see R. Brownsword, J. Wale, *Testing Times Ahead: Non-Invasive Prenatal Testing and the Kind of Community that We Want to Be*, in *Modern Law Review,* 81, 2018, 646.

[58] A. Newson, *Should Parental Refusals of Newborn Screening Be Respected?*, in *Cambridge Quarterly of Healthcare Ethics,* 15, 2, 2006, 135-146.

[59] In the UK, these limits may be influenced by the age, maturity, and capacity of the child (*Gillick v West Norfolk and Wisbech Area Health Authority* [1986] AC 112; *Re D (A Child)* [2019] UKSC 42). At international level, there is legal recognition that interventions against individuals without capacity to consent must generally be undertaken for their direct benefit: *Oviedo Convention on Human Rights and Biomedicine*, Council of Europe Treaty no 164, Art 6(1).

and prohibition can be general, applying to all human persons,[60] and addressing specific categories of person (eg minors)[61] or relationships.[62] Commonly there will be legal frameworks regulating the acquisition of legal responsibility and authority for a minor;[63] and, wardship and care proceedings can restrict or qualify the rights and responsibilities of natural parents over their children.

What is more controversial is the nature and extent of the obligations that parents owe to their children in terms of general nurture, environment, and life choices. There has been much discussion around whether children have a right to an open future,[64] and what, if any, correlative parental obligations there might be: for example, are there parental duties to preserve or not close down future options for children?[65] In the seminal case of *Gillick*, Lord Scarman claimed that the purpose of parental power and control over the person and property of a child: "exists primarily to enable the parent to discharge his duty of maintenance, protection, and education until he reaches such an age as to be able to look after himself and make his own decisions".[66]

Thus envisaged, a parent can consent or withhold agreement to NBS in discharge of their duty to maintain or protect, with moral limits or boundaries that are capable of extending beyond any legal constraint or interference by the State. This is important because NBS is capable of yielding information that could compromise or impede the future life options of the test subject – for example, a genetic disorder that may never become symptomatic which is nonetheless a condition to be disclosed for insurance or employment purposes later in life. Framing parental consent as necessary to the extent that it serves the interests of the newborn, offers a platform to circumvent or qualify the sovereignty of consent. However, if the question is about the interests of a baby, rather than about proxy consent as such, a new point of dispute is introduced into the relationship between parents and healthcare professionals.

3.4.2. Competing and conflicting interests

Where parental interests are set against societal interests, the issue is often framed in terms of private against public interest, with the former operating as an "effective discussion stopper"[67] against those advancing wider societal claims. However, the relationship between the public and private is unstable and there are overlaps between these domains that make this division potentially problematic. The issue is therefore not simple recognition of the existence of a private or public interest in screening newborns, rather it is determining which interests – particularly interests in autonomy and in informed consent – are legitimate in the circumstances, and where the balance of interests and resulting priorities rest.

[60] For our purposes, born humans beings that qualify for legal protection. We accept that there might be qualified protection for those beings in persistent vegetative/ brain dead states (*Bland v Airedale NHS Trust* [1993] AC 789).

[61] Sexual Offences Act 2003.

[62] *Ibid*, S16.

[63] Children Act 1989 (as amended).

[64] J. FEINBERG, *The Child's Right to an Open Future*, in *Freedom and Fulfillment*, Princeton, 1992.

[65] J. WALE, *Regulating disruptive technology and informational interests in the arena of reproductive tests,* cit.

[66] *Gillick v West Norfolk and Wisbech Area Health Authority*, cit., para 185E.

[67] S. VAN DER BURG, A. OERLEMANS, *Fostering caring relationships: Suggestions to rethink liberal perspectives on the ethics of newborn screening*, cit., 181.

To start with the interest in autonomy, even if it is agreed that parents have an interest in making *their own* choices, or in a "sphere of decision privacy",[68] this leaves open to debate the range of choices or decisions that should be available to parents. Moreover, because newborn infants do not have the immediate capacity to make their own choices, this gives the State a potential foothold in decision-making – for example, it might be argued that concerns about vaccination, education or other significant health risks that might cause serious harm to a child, justify the use of the *Parens Patriae* jurisdiction to override any parental refusal or action.[69] That is not to say that the Courts will necessarily discount parental views, but it does mean that it is far easier for State actors to claim a legitimate interest in decisions that concern those without capacity to act and make choices independently.

Nevertheless, is it legitimate for the State to compel acts that are designed to protect or prevent possible serious harm to a child, even if that child is asymptomatic? A claim might be made that NBS serves a moral imperative or moral responsibility to ensure the health of natural children[70] – but this is surely doubtful when using whole exome or genome sequencing. Short of compulsion, the State may influence practices by encouragement, nudges and incentives around NBS.[71] That said, at all levels, State actors need to be mindful that NBS programmes need parental support and endorsement to be effective, and policymakers alienate parents at their risk. Accordingly, healthcare professionals have to balance the need for parental trust, harm avoidance and the delivery of beneficent outcomes for test subjects.[72]

Against the idea of a parental *right* to autonomy, the emphasis is sometimes on "duty". Indeed, in the seminal case of *Montgomery v Lanarkshire Health Board,*[73] the primary concern of the court in the negligence action was whether the treating doctor had complied with the duty owed to the pregnant patient in terms of informational exchange. These references to the duties of doctors could be simply the other side of patients' rights; but, they might also betray an adjustment to the scope or weight of the interest and, in effect, a downgrading of the right.[74]

Turning to the parents' interest in being informed, it was recognised in *Montgomery* that "the doctor's duty is not fulfilled by bombarding the patient with technical information which she cannot reasonably be expected to grasp, let alone by routinely demanding her signature on a consent

[68] Compare J. WILSON, *Is respect for autonomy defensible?*, in *Journal of Medical Ethics,* 33, 2007, 353.

[69] See for example, *Re C (A child) (HIV testing)* [2000] 1 WLR 2. *Parens Patriae* is the authority of the State to protect those that are unable to protect themselves. International legal instruments (eg. the *Oviedo Convention*, Art 6(1)) might prefer to frame the intervention in terms of potential benefit rather than harm.

[70] See for eg., F. ULPH, N. DHARNI, R. BENNETT, T. LAVENDER, *Consent for newborn screening: screening professionals' and parents' views*, cit., 155.

[71] A. NEWSON, *Should Parental Refusals of Newborn Screening Be Respected?,* cit.

[72] S. VAN DER BURG, A. OERLEMANS, *Fostering caring relationships: Suggestions to rethink liberal perspectives on the ethics of newborn screening*, cit., 180.

[73] *Montgomery v Lanarkshire Health Board* [2015] UKSC 11.

[74] See, further, B.J. RICHARDS, *Autonomy and the Law: Widely Used, Poorly Defined,* in D.G. KIRCHHOFFER, B.J. RICHARDS (eds), *Beyond Autonomy: Limits and Alternatives to Informed Consent in Research Ethics and Law*, Cambridge, 2019; and, M. DUNN, K.W.M. FULFORD, J. HERRING et al., *Between the Reasonable and the Particular: Deflating Autonomy in the Legal Regulation of Informed Consent to Medical Treatment,* in *Health Care Analysis,* 27, 2018, 110.

form".[75] Informational transfer on its own is insufficient and there needs to be some level of patient understanding and knowledge about the available choices, risks etc.[76] Whether viewed as a right or as a duty, the challenges associated with securing participant understanding are amplified where genomics are involved.[77] Exactly how far healthcare professionals need to go to unearth understanding and values before seeking consent and undertaking invasive investigation or treatment remains unclear. What is reasonably clear is that informed consent processes may have a value if they facilitate and evidence the knowledge and understanding of those who provide consent. Mere written agreement or verbal affirmation to the investigation or interference is unlikely to meet these objectives.

While some might jib at the sovereignty of consent, thinking that it overplays the legitimate interests of parents, there is more likely to be assent to the sufficiency of consent and to there being at least a prima facie case for parents playing a central role in NBS decision-making. There are also good and independent reasons why many screeners, even if they are not dyed-in-the-wool rights theorists, might want to insist on a demanding threshold for a valid consent – by seeking a free and informed consent, healthcare professionals signal respect, maintain trust and help avoid legal disputes.[78] Certainly, where a community takes rights seriously, the burden should be on those who want to either de-centre or dilute consent to justify such modification or relaxation.

3.4.3. Taking stock

From the many particular points made in the course of our discussion, there are perhaps four key points to highlight. First, even in ordinary times, biolegal and bioethical debates do not stand still. We now live in an age of genomics (and other omics) all creating new opportunities but also challenges for the governance of NBS. Screening today is not screening as Wilson and Jungner knew it. Secondly, for at least two reasons, mapping and classifying NBS regimes relative to our ideal-typical model of consent is not straightforward. One reason is that there can be some distance between the promise of screening regimes and their actual practice. Another reason is that NBS programmes involve a number of elements each of which, in principle, could be subject to parental consent. So, mandatory regimes that do not require parental consent in relation to the basic elements of NBS and the storing of blood might make consent a requirement in relation to other elements (such as the return of findings). Thirdly, the complexity of genetic data means that, even if the sovereignty of consent is not questioned, even if there is no resistance to parental rights – admittedly, an unlikely scenario in biolaw and bioethics as we have come to know them in ordinary times – the implementation of the ideal-typical model of consent will be a major challenge. Fourthly, while we can identify the four critical points at which pressure might be applied to a rights-based regime and parental consent, we cannot always anticipate from which ethical constituency pressure will be applied any more than we

[75] *Montgomery v Lanarkshire Health Board*, at para 90 (Lords Kerr and Reed).
[76] B.J. RICHARDS, *Autonomy and the Law: Widely Used, Poorly Defined*, cit., 30.
[77] N. ASHLEY, M.A. TOMLINSON, D. SKINNER et al., *"Not tied up neatly with a bow": Professionals' Challenging Cases in Informed Consent for Genomic Sequencing*, in *Journal of Genetic Counseling*, 25, 1, 2016, 62-72, doi:10.1007/s10897-015-9842-8.
[78] F. ULPH, S. WRIGHT, N. DHARNI, et al., *Provision of Information about newborn screening antenatally: a sequential exploratory mixed-methods project,* cit.

can anticipate where pressure will be applied or the form that it will take – whether it is to displace or downgrade rights or to de-centre or dilute consent. Furthermore, predicting the outcomes of debates and disputes around the proposed expansion of NBS is not easy. In the cut and thrust of these debates, our ideal-typical model of consent and the rights on which it is based might or might not hold their ground.

4. In Extraordinary Times: Proxy Consent and Pandemics

From ordinary times, we turn to extraordinary times. In ordinary times, the pattern of debate in biolaw and bioethics, although pluralistic and contested, is reasonably familiar; the conversation is one we know. In extraordinary times, there is a different conversation, one that might employ familiar ideas but one that appeals to justifications that are far from ordinary.

If recent experience with CoViD-19 is representative of the way in which communities (local, regional, and international) reason during the time of a pandemic, then it is pretty clear that the case for public health measures does not rest on the consent of individuals. It is pretty clear, in other words, that if parents were to try to stand on their rights and resist, say, the vaccination of their children or some other harm-reducing measure advised by public health professionals, there would be a major push-back[79]. Similarly, we would expect there to be a push-back if parents were to resist or refuse to consent to the participation of their babies or children in non-invasive studies that epidemiologists believe would illuminate our understanding of the nature of the pandemic-causing virus. In this part of the article we sketch the changing rhetoric; we relate this to three possible accounts that purport to explain and to justify the displacement of rights and consent; and then we suggest that one of these accounts, engaging the idea of a stewardship responsibility for the global commons,[80] offers the best account of what makes any particular time or issue "extraordinary" and, with that, offers the best understanding of the extent to which rights and consent are justifiably displaced.

4.1. Rights and Consent Superseded

In extraordinary times, even in a community of rights, consent is not everything. Alongside ordinary times conversations and contestation, there are new priorities and a sense that we are now operating beyond both biolaw and bioethics as we ordinarily know them.

[79] That said, to a certain extent, this is context-sensitive. For example, a recent poll in the USA, suggests that about a third of US adults would decline a vaccine for CoViD-19 and, presumably, these adults would also push-back against mandatory vaccination of their children: see S.M. O'KEEFE, *One in Three Americans Would Not Get COVID-19 Vaccine*, August 7, 2020, available at https://news.gallup.com/poll/317018/one-three-americans-not-covid-vaccine.aspx (last accessed August 28, 2020).

[80] Compare, Nuffield Council on Bioethics, *Public health: ethical issues*, November 2007. The Council's reliance on the concept of stewardship attracted some criticism as being (from a utilitarian perspective) unnecessary. However, provided that stewardship is understood as operating in a different (extraordinary times) domain from ordinary time utilitarian reasoning, it is an evocative and defensible idea. For defence, see T. BALDWIN, R. BROWNSWORD, H. SCHMIDT, *Stewardship, Paternalism and Public Health: Further Thoughts,* in *Public Health Ethics,* 1, 2009, and R. BROWNSWORD, *Regulation: Prudence, Precaution and Stewardship*, in *Northern Ireland Legal Quarterly,* 62, 2011, 573.

If this meant that, in extraordinary times, there are no longer legal or ethical constraints or that we should submit to a Leviathan, this would be deeply worrying. However, this is not the case: there remains a conspicuous concern to do the right thing. For example, the European Group on Ethics in Science and New Technologies (EGE) concludes its *Statement on European Solidarity and the Protection of Fundamental Rights in the COVID-19 Pandemic* with the following ringing declaration: "[w]e must live through this pandemic, and after it. We must face this situation with strength, care and solidarity – a social vaccine that accompanies our search for a CoViD-19 vaccine, which has an enduring character. One that provides resilience, lasting social and economic solidarity and *lasting immunity against indifference*".[81]

Nor is it the case that rights-based thinking is displaced from conversations that bring ordinary times values to bear on governance in extraordinary times. Indeed, the EGE, echoing the concerns of civil libertarians, insists that the unprecedented quarantine measures that were adopted to confine the spread of the virus, the extended use of surveillance technologies, and the like, should respect human rights by being no more than necessary and proportionate. As the EGE says, "[t]he public health emergency must not be abused to usurp power, or to permanently suspend the protection of rights and liberties".[82] This is also not to say that concerns about consent are altogether set aside – for example, if children or their parents were to be conscripted into research trials, or if post-mortem samples were to be taken by researchers without consultation with families, it would be no surprise at all if consent were to re-surface as a basic requirement.[83] Nevertheless, the dominant thoughts provoked by a pandemic are about taking measures that, all things considered, will be for the benefit of human health and well-being, about keeping people safe, about reducing avoidable pressure on the healthcare infrastructure, and about maintaining social solidarity.

This shift in thinking prompted by pandemics implies that, in some circumstances, where communities are faced by emergencies or catastrophes, neither the consent of individuals nor their particular rights are central to our justificatory thinking.[84] Instead, there is a renewed emphasis on collective well-being and responsibility. Reflecting this shift, in its *Statement on COVID-19: Ethical Considerations from a Global Perspective*, the UNESCO International Bioethics Committee says that the responsibilities include those of "governments to ensure public safety and protect health, and raise awareness of the public and other actors on the methods required for this purpose; responsibilities of the public to abide by the rules that protect everyone not only as individuals but also, and above all, as a community; [and] responsibilities of healthcare workers to treat and care for patients".[85]

[81] Statement issued April 2, 2020, p 4, emphasis supplied. Available at https://bit.ly/3apLyMM (last accessed July 7, 2020).

[82] Recommendation 4. Compare M.M. MELLO, C.J. WANG, *Ethics and governance for digital disease surveillance*, in *Science,* 368, 6494, 2020, 951.

[83] Compare K. MOODLEY, B.W. ALLWOOD, T.M. ROSSOUW, *Consent for critical care research after death from COVID-19: Arguments for a waiver*, *South African Medical Journal,* 2020, 629-634.

[84] That said, the idea of collective consent (which, of course, departs from our ideal-type) might play a role in weakening the significance of individual rights and personal consent.

[85] SHS/IBC-COMEST/COVID-19, Paris, 26 March 2020.

The question then is this: in which circumstances are there compelling reasons for refocusing our justificatory reason in this way? How should we account for the displacement of rights and consent?

4.2. Three Accounts of the Displacement of Rights and Consent

We suggest that there are three principal narratives that might both explain and defend the displacement, in exceptional times, of rights and consent. Here, we will simply sketch the three accounts and then we will elaborate on the third narrative in the next sub-section of the article.

The first narrative is along the lines that the scale and immediacy of the threat represented by a pandemic takes us into a "state of exception". In these exceptional circumstances, ordinary bioethics is suspended. The imperative is to respond to the threat in a way that will prevent the spread of the infection and mitigate its harmful effects. Basically, governments and public health agencies must do whatever it takes. As we have already said, the rhetoric around CoViD-19 does not entirely fit with this narrative and, more importantly, to give "the authorities" a licence of this kind is a hostage to fortune – if "states of exception" are to be recognised, they need to be for no longer than absolutely necessary.

The second narrative treats a pandemic as a radical change to the context in which bioethics is ordinarily conducted but it resists the idea that bioethics is now suspended and superseded by a state of exception. To the contrary, bioethics as practised in ordinary times now extends into extraordinary times, but the pattern of advantage and disadvantage alters because of the radical change in context. Whereas, in ordinary times and ordinary contexts, the ethic of rights and consent is an important voice in bioethical debate, in extraordinary times and extraordinary contexts, this voice loses its power and influence. In other words, our attraction and commitment to an ethic of rights and consent is context-dependent; and, in some exceptional circumstances, that attraction and commitment weakens.

The third narrative rejects the idea that a pandemic introduces a state of exception in the sense of a licence for governments to do whatever it takes; and it also rejects the idea that the pandemic simply changes the context in which ordinary times bioethical contestation takes place. Rather, the third narrative identifies the pandemic as a particular threat to the conditions which make it possible to adopt bioethical positions and to engage in bioethical arguments in the first place. In the third account, we are reminded not only of the vulnerability of humans and the fragility of the global commons on which all forms of human social existence depend, but also that, while some threats to the commons are *acute* (as is the case with a pandemic), others are *chronic* and *incremental* (as is the case, for example, with climate change and with big data and surveillance). By contrast with the first two accounts, in which there is a period of ordinary times, then a period of extraordinary times before a return to ordinary times, the third account holds that, even in what are ostensibly ordinary times with their standard debates, there needs to be the kind of monitoring, vigilance, and precautionary preparedness that is appropriate in extraordinary times.

Following this third account, ordinary time conversations and ordinary case justifications co-exist with what we are calling extraordinary time conversations and stewardship justifications. In this bigger picture, where stewardship justifications are brought into play (whether by acute or by chronic threats) it is not a case of the triumph of one kind of ordinary case justification, it is not the

values of one community (where neither rights nor consent are taken seriously) displacing the values of another community (where rights and consent are taken seriously), but a case of stewardship responsibilities for the global commons supervening on the values of all communities.

4.3. Stewardship and the Global Commons

The global commons has two dimensions: one relates to human existence; and the other relates to the human capacity for agency. In the case of a pandemic such as CoViD-19, there is an urgent need not only to protect the conditions for human life but also to minimise the compromising of the context for agency.

First, the *human* species is defined by its biology; and the prospects for human life depend on whether the conditions are compatible with the biological characteristics and needs of the *human* species. Most planets will not support *human* life. The conditions on planet Earth, neither too hot nor too cold, are special for *humans*. However, the conditions are not specially tailored to the needs of any particular human; these are the generic conditions for the existence of any member of the human species.

Secondly, it is characteristic of human *agents* that they have the capacity to choose and to pursue various projects and plans whether as individuals, in partnerships, in groups, or in whole communities. Sometimes, the various projects and plans that they pursue will be harmonious; but, often, human agents will find themselves in conflict or competition with one another. However, before we get to conflict or competition, there needs to be a context in which the exercise of agency is possible. This context is not one that privileges a particular articulation of agency; it is prior to, and entirely neutral between, the particular plans and projects that agents individually favour; the conditions that make up this context are generic to agency itself.

Any human agent, reflecting on the antecedent and essential nature of the commons must regard the critical infrastructural conditions as special. From any practical viewpoint, prudential or moral, that of regulator or regulatee, the protection of the commons must be the highest priority.[86] Protective stewardship will be guided by three imperatives.

In the first instance, it is imperative that steps are taken to protect, preserve and promote the natural ecosystem for human life.[87] At minimum, this entails that the physical well-being of humans must be secured; humans need oxygen, they need food and water, they need shelter, they need protection against contagious diseases, if they are sick they need whatever medical treatment is available, and they need to be protected against assaults by other humans or non-human beings.

The second imperative is to construct and maintain the conditions for meaningful self-development and agency: there needs to be a sufficient sense of self and of self-esteem, as well as sufficient trust

[86] An understanding of what it is to have the capacity for agency presupposes respect for the conditions for both self-interested agency and other-regarding agency. To cash out this argument, see A. GEWIRTH, *Reason and Morality*, Chicago, 1978; D. BEYLEVELD, *The Dialectical Necessity of Morality,* Chicago, 1991; and ID., *What Is Gewirth and What Is Beyleveld: A Retrospect with Comments on the Contributions*, in P. CAPPS, S.D. PATTINSON (eds), *Ethical Rationalism and the Law,* Oxford, 2017, 233.

[87] Compare, J. ROCKSTRÖM et al., *Planetary Boundaries: Exploring the Safe Operating Space for Humanity*, in *Ecology and Society*, 14, 2009, 32, available at: http://www.ecologyandsociety.org/vol14/iss2/art32/ (last accessed November 14, 2016); and, K. RAWORTH, *Doughnut Economics*, London, 2017, 43-53.

and confidence in one's fellow agents, together with sufficient predictability to plan, so as to operate in a way that is interactive and purposeful rather than merely defensive. The context should support agents in being able to freely choose their own ends, goals, purposes and so on ("to do their own thing") as well as to form a sense of their own interests and identity ("being their own person").[88] With existence secured, and under the right conditions, human life becomes an opportunity for agents to be who they want to be, to have the projects that they want to have, to form the relationships that they want, to pursue the interests that they choose to have and so on.

Thirdly, the commons must secure the conditions for an aspirant moral community, whether the particular community is guided by teleological or deontological standards, by rights or by duties, by communitarian or liberal or libertarian values, by virtue ethics, and so on. The generic context for moral community is impartial between competing moral visions, values, and ideals; but it must be conducive to "moral" development and "moral" agency in a formal sense. In particular, moral community of any kind presupposes a context in which agents are free to form and then to act on their own judgments of what it is to do the right thing.[89]

While respect for the commons' conditions is binding on all human agents, it should be emphasised that this does not rule out the possibility of prudential or moral pluralism. Rather, the commons represents the pre-conditions for both individual self-development and community debate, giving agents and communities the opportunity to develop their own view of what is prudent as well as what should be morally prohibited, permitted, or required. Whether the issue is the extension of NBS, or responding to a pandemic, or any other matter, it is the commons that provides the platform for such reflection, development, and debate.

5. Conclusion

This article has highlighted the challenges facing parents who, acting as proxies for their children, seek to stand on their rights and the need for their consent. Disputes around NBS are a case in point. Even in what we are calling ordinary times, the rights of parents and their giving or withholding of consent can come under increased pressure as developments in genetics offer reasons for undertaking more extensive screening. While such pressure might not displace rights, the conditions for a valid consent might be diluted. In what we are calling extraordinary times, the arguments against proxy consent become overwhelming: neither individual rights nor informed consent are now focal. Rather, it is our responsibilities as stewards of the commons that becomes the key justificatory consideration.

[88] Compare the insightful analysis of the importance of such conditions in M. BRINCKER, *Privacy in Public and the Contextual Conditions of Agency,* in T. TIMAN, B.C. NEWELL, B.-J. KOOPS (eds), *Privacy in Public Space*, Cheltenham, 2017, 64; and, similarly, see M. HU, *Orwell's 1984 and a Fourth Amendment Cybersurveillance Nonintrusion Test, Washington Law Review,* 92, 2017, 1819, at 1903-1904.

[89] See, e.g., R. BROWNSWORD, *Code, Control, and Choice: Why East is East and West is West,* in *Legal Studies,* 25, 2005, 1; ID., *So What Does the World Need Now? Reflections on Regulating Technologies,* in R. BROWNSWORD, K. YEUNG (eds), *Regulating Technologies,* Oxford, 2008, 23; ID., *Lost in Translation: Legality, Regulatory Margins, and Technological Management,* in *Berkeley Technology Law Journal,* 26, 2011, 1321.

The juxtaposition of ordinary time justifications with extraordinary time justifications opens an agenda for further inquiry. In particular, further reflection is invited on when the extraordinary is engaged and when it is not; on the character of extraordinary time reason, and any "reach-through" from one class of justifications to the other; and, on how to operationalise stewardship in extraordinary times.

With regard to the first question, we want to have the right justificatory conversation at the right time. To do this, we need to be clear about whether a particular question is appropriately treated as an ordinary times matter or whether it engages extraordinary considerations. For example, we have treated debates about the extension of NBS as an ordinary time matter – and we have no reason to think otherwise. However, in the context of a pandemic, some aspects of NBS might assume significance relative to mitigating the risks presented by the virus in which case this becomes a matter for extraordinary time justifications. Without such clarity, we might continue to rely on ordinary time justifications when the commons is already being compromised (as, for example, some might argue is the case with climate change); and, conversely, we might continue to rely on extraordinary time justifications when ordinary time considerations should be applied (as is the fear of civil libertarians about the persistence of restrictions imposed at the height of, and in the wake of, CoViD-19). In this light, we should be careful with narratives that imply that we live through a linear sequence of periods (ordinary, then extraordinary, then back to ordinary); rather, as the third narrative implies, we now live through a period in which the questions that we debate and the challenges that we face sometimes engage, as it were, ordinary time considerations but sometimes (as with climate change and pandemics) extraordinary time considerations.

Secondly, there are questions about the character of supervening reason and whether there is any reach-through of ordinary time values and justifications to extraordinary times. In ordinary times, we differentiate between prudential reason and moral reason. However, commons' protecting reason seems to be both prudential and moral; it is in the interest of everyone and it is categorical, exclusionary, and overriding. Beyond this particular question of character, as we have said, even in extraordinary times, some familiar ordinary times values will resurface in some debates (debates that actually belong to ordinary times). However, the question is whether values of this kind reach through to the stewardship of the commons. For example, some might argue that values such as autonomy, privacy, and human dignity are fundamental not only to the constitution of a community of rights but also to the context for agency that is one of the dimensions of the commons' conditions.

A third question, vividly highlighted by the recent experience with CoViD-19, is about the coordination of our stewardship responsibilities. In principle, we are all stewards for the global commons and, as such, we can "do our bit" – for example, we can comply with the necessary restrictions on our movement or association that are put in place to prevent the spread of the virus. However, in practice, the restoration and maintenance of the global commons needs international leadership.[90] In the case of a pandemic, it is the WHO that is the obvious candidate. However, if the

[90] See R. BROWNSWORD, *Redesigning the Institutional Framework II: International Institutions*, in ID., *Law 3.0: Rules, Regulation and Technology*, Abingdon, 2020.

ISSN 2284-4503

WHO is to be hobbled and undermined by great powers that conduct international relations in an entirely self-serving nationalistic way, there has to be some other approach.[91]

Finally, we might also reflect on the more general jurisprudential implications of the co-existence of ordinary time and extraordinary time justifications. While we might be more familiar with the former, it is in the latter that we have real terra firma for our justificatory arguments in biolaw and bioethics. If, as Sarah Franklin has argued, biolaw and bioethics have lost their bearings, then it is with our responsibilities in relation to the global commons that we should begin the work of restoration.[92] After all, unlike ordinary time debates where people find it hard to agree, no one should find it hard to agree that we should take special care of conditions that are neutral between humans, neutral between articulations of self-interest, and neutral between articulations of a moral viewpoint but without which humans cannot exist, cannot form a sense of their self-interest, and cannot exercise moral agency.

[91] See A. JOSEPH, H. BRANSWELL, *Trump: US will terminate relationship with the World Health Organization in wake of Covid-19 pandemic*, in *STAT*, May 29, 2020, available at https://bit.ly/3tGkW1B (last accessed, July 5, 2020).

[92] S. FRANKLIN, *Ethical research – the long and bumpy road from shirked to shared.* in *Nature,* 574, 2019, 627-630, doi: 10.1038/d41586-019-03270-4.

Venire senza *provenire:* il diritto di accesso dell'adottato alle proprie origini nelle strettoie dell'anonimato materno

Stefano Agosta*

COMING WITHOUT *COMING FROM*: THE ADOPTEE'S RIGHT OF ACCESS TO ORIGINS WITHIN THE CONSTRAINTS OF MATERNAL ANONYMITY

ABSTRACT: This article analyses the judicial path, crossed by lights and shadows, and its ability to make it feasible to more easily learn information about one's own parental history in anonymous birth. A comparison is made between the European Court of Human Rights (ECtHR) case law and the Italian Constitutional Court case law, showing differences in the methods but strong similarities in the substantive solutions. Conclusively, in the Italian legal system, the mother's decision to confirm her original choice for anonymity has an undisputed prevalence when it tries to balance with the child's constitutional right to have his or her own personal experience recognised.

KEYWORDS: Anonymous birth; right to know one's origins; right to respect for private life

SOMMARIO: 1. Identità personale e conoscenza delle origini: premessa – 2. Diritto di *non farsi trovare* (della madre) e diritto di *ricercare* (dell'adottato) a confronto: nella prospettiva *statica* – 3. (*Segue*): nel mosaico costituzionale (tra salute, vita privata e identità individuale) – 4. (*Segue*): nella dimensione *dinamica* – 5. Centralità del veto materno e residue ambiguità di un bilanciamento (mascherato).

1. Identità personale e conoscenza delle origini: premessa

Tra le non poche intersezioni al presente inevitabilmente esistenti – per facilmente intuibili ragioni legate anche al progressivo avanzamento del progresso medico-scientifico – tra diritto e genetica, un cospicuo posto in giurisprudenza e nella dottrina se l'è senz'altro ritagliato nel corso degli ultimi anni (e continua con forza a detenerlo) il diritto della persona alla ricerca delle proprie origini genetiche e biologiche.

Se perlomeno due sono gli angoli visuali da cui la delicata questione della conoscenza della propria storia personale nel nostro ordinamento può essere complessivamente riguardata – e cioè, rispettivamente, dalla prospettiva, per così dire, dei *poteri* ovvero da quella dei *diritti* – in questa sede un'attenzione particolare sarà riservata solo al secondo dei due richiamati profili[1], con specifico riferimento al diritto all'identità personale *ex* all'art. 2 Cost. di cui l'esigenza di accedere al proprio pre-

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[1] … con la riserva naturalmente di approfondire in un'altra occasione il primo dei due aspetti considerati.

gresso vissuto non può che rappresentare uno dei più salienti aspetti[2]. Non rappresenta, d'altro canto, un mistero che «lo sviluppo equilibrato della personalità individuale e relazionale» di ciascuno, per un verso, passa dalla «costruzione della propria identità esteriore, di cui il nome e la discendenza giuridicamente rilevante e riconoscibile costituiscono elementi essenziali» ma, per un altro, non può parimenti prescindere dalla edificazione di una speculare identità «interiore» appunto richiedente «la conoscenza e l'accettazione della discendenza biologica e della rete parentale più prossima»[3].
Da questo punto di vista, è perciò ovvio che la medesima vita relazionale dell'individuo non possa che profondamente risentire del corretto appagamento o meno dell'innata esigenza di ciascuno di apprendere le informazioni relative alla propria precedente storia parentale[4]. Certamente valevole per la condizione dell'adottato (cui il presente contributo è espressamente dedicato), tale affermazione pare tanto più vera per altre situazioni alla prima in qualche misura assimilabili come, ad esempio, quella del nato da inseminazione eterologa[5] o a ritroso (ed *in limine*) degli embrioni soprannumerari e potenzialmente destinabili alla c.d. adozione per nascita[6].

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[2] In tal senso, Corte cost. sent. n. 286/2016 (punto 3.4.1. *cons. dir.*, primo cpv) su cui, *ex multis*, E. MALFATTI, *Illegittimità dell'automatismo, nell'attribuzione del cognome paterno: la "cornice" (giurisprudenziale europea) non fa il quadro*, in *forum costituzionale,* 5 gennaio 2017; S. SCAGLIARINI, *Dubbie certezze e sicure incertezze in tema di cognome dei figli*, in *rivista AIC*, 19 maggio 2017, disponibile in https://bit.ly/2ReyFyF; C. INGENITO, *L'epilogo dell'automatica attribuzione del cognome paterno al figlio (Nota a Corte costituzionale n. 286/2016* ed A. FUSCO, *«Chi fuor li maggior tui?»: la nuova risposta del Giudice delle leggi alla questione sull'attribuzione automatica del cognome paterno. Riflessioni a margine di C. cost. sent. n. 286 del 2016*, entrambe in *osservatorio AIC*, rispettivamente, 31 maggio e 5 settembre 2017, disponibili ai seguenti link: https://bit.ly/3o7hs6o e https://bit.ly/3hmtg3g.

[3] In questa direzione, Corte cassaz., sez. I civ., sent. 29 maggio 2017-20 marzo 2018, n. 6963 (rispettivamente, punto 8 e 8.1, primo cpv, *cons. dir.*) su cui, tra i tanti, G. VASSALLO, *Parto anonimo: diritto di conoscere le proprie origini va esteso alle sorelle*, in *altalex,* 12 aprile 2018; E. CATALANO, *Il diritto alla conoscenza delle proprie origini*, in *salvisjuribus*, 4 luglio 2018; A. GIURLANDA, *Il diritto a conoscere le proprie origini può essere esercitato anche nei confronti delle sorelle e dei fratelli biologici dell'adottato?*, in *questione giustizia,* 26 settembre 2018; G. CASABURI, *Riflessioni estemporanee su azioni di stato, nuova genitorialità, tutela del minore,* en attendant *le SS.UU. del 6 novembre 2018*, in *articolo29*, 8 novembre 2018; C. GRANATA, *Il diritto alla ricerca delle proprie origini: i punti rimasti irrisolti dopo la sentenza n. 6963 della Corte di Cassazione, Sez. I, del 20.03.2018*, in *rivista camminodiritto,* 16 dicembre 2019; I. LOMBARDINI, *Il procedimento di "interpello" della madre biologica, che abbia dichiarato di non voler essere nominata al momento del parto, ai fini dell'eventuale revoca dell'originaria dichiarazione, e la progressiva espansione del diritto dell'adottato alla conoscenza delle proprie origini biologiche ad opera della recente giurisprudenza*, in *diritto,* 5 giugno 2020.

[4] Così, Corte cost. sent. n. 278/2013 (punto 4 *cons. dir.*, ottavo cpv) su cui, *ex plurimis*, E. FRONTONI, *Il diritto del figlio a conoscere le proprie origini tra Corte EDU e Corte costituzionale. Nota a prima lettura sul mancato ricorso all'art. 117, primo comma, Cost., nella sentenza della Corte costituzionale n. 278 del 2013* e A. RAPPOSELLI, *Illegittimità costituzionale dichiarata ma non rimossa: un "nuovo" tipo di sentenze additive?*, entrambi in *osservatorio aic,* rispettivamente, dicembre 2013 e gennaio 2015, disponibili ai seguenti link: https://bit.ly/2R2FaEX e https://bit.ly/2Qajbvh.

[5] «In questa dimensione personale», difatti, «le possibilità offerte dalle tecniche di PMA, detta 'eterologa', risolvono problemi medici, ma modificano nel profondo la genitorialità e complicano le domande sulla ricerca delle proprie origini»: in tal senso, ad esempio, V. DE SANTIS, *Diritto a conoscere le proprie origini come aspetto della relazione materna. Adozione, PMA eterologa e cognome materno*, in *nomos – le attualità nel diritto*, marzo 2018, spec. 1. D'altro canto, sarebbe proprio «l'entrata in scena dei "donatori'"» a porre «la grave questione del diritto umano alla conoscenza della proprie origini, questione che non può essere trascurata, minimizzata o schiacciata dal peso degli aspetti tecnici e sanitari»: così, M. CASINI, C. CASINI, *Il dibattito sulla PMA eterologa*

Quale peculiare aspetto del più ampio e sfaccettato diritto all'identità personale – per tornare alla specifica situazione dell'adozione – è perciò ovvio che quello alla ricerca delle proprie origini abbia nel corso degli ultimi anni beneficiato di un più o meno ampio riconoscimento sia a livello internazionale che, per quanto più da vicino ci riguarda, domestico. Sofferto e non poco travagliato frutto di una serie di progressive aperture andate gradatamente (ma non sempre ordinatamente…) stratificandosi nel tempo[7], la disciplina italiana è nondimeno approdata, infine, all'oltremodo severo esito di negare all'adottato qualsivoglia autorizzazione a conoscere la propria storia personale – e a chiunque vi abbia interesse, se non siano prima trascorsi ben cento anni dalla formazione del certificato di assistenza al parto ovvero della cartella clinica riportanti i dati identificativi della partoriente – senza neppure contemplare una previa verifica della persistente volontà materna di rimanere nell'anonimato.

In presenza di talmente stringenti condizioni peculiarmente dettate dal diritto *vigente* legislativo era perciò prevedibile (se non, persino, inevitabile) che sulle tracce di un più ragionevole bilanciamento tra gli interessi coinvolti – «quello della persona che vuole completare la costruzione della propria identità attraverso la ricerca delle origini biologiche e quello della madre biologica che ha esercitato, al momento del parto, il diritto di non essere nominata e che può voler conservare questo segreto proprio al fine di non alterare l'identità anche relazionale costruita nel tempo»[8] – si sarebbe ben presto messo pure, se non soprattutto, il diritto *vivente* giurisprudenziale: facendo logicamente-cronologicamente precedere alla disamina in senso *dinamico* delle istanze costituzionali in campo in-

all'indomani della sentenza costituzionale n. 162 del 2014. In particolare: il diritto a conoscere le proprie origini e l'"adozione per la nascita", in *BioLaw Journal*, 2, 2014, 139.

[6] Sulla gigantesca questione non è, com'è ovvio, possibile adesso diffusamente soffermarsi se non appena per rilevare come già «nell'ambito della discussione sulla legge 40/2004, fu proposta l'"adozione per la nascita" o "adozione prenatale" come rimedio transitorio, limitato e temporaneo, sul presupposto che con l'entrata in vigore della nuova legge, sarebbe cessato l'accumulo di embrioni di scorta nei congelatori»: duramente bocciata, infine, da quanti «vedevano nella "dichiarazione di adottabilità del concepito" la equiparazione dei non nati ai già nati» (così, nuovamente, M. CASINI-C. CASINI, *op. cit.*, 151), nel corso degli anni successivi tale proposta è poi tornata in auge grazie ad una serie di interventi del Comitato nazionale di bioetica che ne hanno, a più riprese, evidenziato la bontà degli argomenti (cfr. part. *Adozione per la nascita degli embrioni crioconservati e residuali derivanti da procreazione medicalmente assistita (P.M.A.)* e *Destino degli embrioni derivanti da procreazione medicalmente assistita e non più impiantabili*, entrambi in http://bioetica.governo.it, rispettivamente 18 novembre 2005 e 26 ottobre 2007). In oggetto, comunque, v. ad esempio A. PALAZZO, *La filiazione*, Milano, 2007, part. 52 ss.; M. PICOZZI, F. NICOLI, V. VIGANÒ, *Il dono tra desiderio e ragione. Una riflessione sui principali nodi bioetici connessi alla fecondazione eterologa*, in L. GRION (a cura di), *Cose o persone? Sull'esser figli al tempo dell'eterologa*, Trieste, 2016, spec. 58 ss.; D. CASTELLANO, *Congelamento degli embrioni: un caso e molti problemi*, in *filodiritto,* 15 dicembre 2020.

[7] In tal senso, cfr. l'originaria formulazione di cui all'art. 28, l. n. 184/1983, *Diritto del minore ad una famiglia*, nonché le successive novelle introdotte, rispettivamente, dall'art. 30, comma 1 (*Dichiarazione di nascita*), D.P.R. n. 396/2000, *Regolamento per la revisione e la semplificazione dell'ordinamento dello stato civile, a norma dell'articolo 2, comma 12, della legge 15 maggio 1997, n. 127*, dall'art. 24, l. n. 149/2001, *Modifiche alla legge 4 maggio 1983, n. 184, recante «Disciplina dell'adozione e dell'affidamento dei minori», nonché al titolo VIII del libro primo del codice civile* e dall'art. 93, comma 2 (*Certificato di assistenza al parto*), D.Lgs. n. 196/2003, *Codice in materia di protezione dei dati personali*.

[8] Così, nuovamente, Corte cassaz., sez. I civ., sent. 29 maggio 2017-20 marzo 2018, n. 6963 (punto 8.1, *cons. dir.*, rispettivamente, secondo e primo cpv) cui si rinvia anche per il passaggio testuale immediatamente successivo.

tanto un'approfondita ricognizione in senso *statico* del «diritto a conoscere la verità sulla propria storia personale» così come, pure, di «quello a conservare la costruzione preesistente dell'identità propria e dei terzi eventualmente coinvolti».
Lasciando per il momento da parte il profilo meramente *sincronico* dei due diritti evocati in campo (correlato, cioè, alla necessità di garantire a concepito e gestante le migliori condizioni di contesto per la nascita e, per questa via, evitare l'assunzione da parte di quest'ultima di scelte irreversibili[9]) per concentrarsi, adesso, solo su quello *diacronico* (di tutto il tempo successivo alla nascita) dei medesimi, è naturale che «l'impegno sul riconoscimento del diritto a conoscere le proprie origini è stato stimolato, in tempi molto recenti, proprio dalla necessità di trovare una composizione equilibrata tra diritti contrapposti»[10].

2. Diritto di non farsi trovare (della madre) e diritto di ricercare (dell'adottato) a confronto: nella prospettiva statica

Già dai pochi rilievi appena svolti non difficile è intuire come – lungi dall'essere artificiosamente separati *in astratto* – un solido e reciproco condizionamento esista nella *pratica* tra le fondamentali esigenze costituzionali retrostanti ai rispettivi diritti di madri e di figli nel caso in esame[11].
Rispetto alla madre, in particolare, si è voluto normativamente evitare che pure a distanza di molti anni costei fosse chiamata a ritornare sui propri passi – su iniziativa di un figlio sconosciuto e magari già grande – statuendo la natura irreversibile dell'originaria scelta per il parto anonimo[12]: il legislatore dell'epoca aveva, insomma, puntato tutto su quell'inestricabile intreccio esistente tra la titolarità del diritto materno all'anonimato ed il suo esercizio inevitabilmente involgente un'irretrattabile di-

[9] Così, Corte cost., sent. n. 278 cit. (punto 4 *cons. dir.*, quarto cpv, primo per.) laddove, in particolare, richiama il medesimo passaggio contenuto nel proprio precedente in materia di cui alla sent. n. 425/2005 (punto 4 *cons. dir.*, terzo cpv) a commento della quale, *ex multis*, S. MARZUCCHI, *Dei rapporti tra l'identità dell'adottato e la riservatezza del genitore naturale (in margine alla sent. n. 425 del 2005 della Corte costituzionale)*, in *www.associazionedeicostituzionalisti.it*, 6 aprile 2006; S. FAVALLI, *Parto anonimo e diritto a conoscere le proprie origini: un dialogo decennale fra CEDU e Corte Costituzionale italiana*, in *forum costituzionale,* 9 dicembre 2013); B. BARBISAN, *Apprendimento e resistenze nel dialogo fra Corte costituzionale e Corte di Strasburgo: il caso del diritto all'anonimato della madre naturale*, in *diritti comparati*, 9 maggio 2016.
[10] Così, ancora, Corte cassaz., sez. I civ., sent. 29 maggio 2017-20 marzo 2018, n. 6963 (punto 8.1, *cons. dir.*, secondo cpv).
[11] Com'è stato d'altro canto rilevato, da Corte cost., sent. n. 278 cit. (punto 4 *cons. dir.*, primo e secondo cpv), «il tema del diritto all'anonimato della madre e quello del diritto del figlio a conoscere le proprie origini ai fini della tutela dei suoi diritti fondamentali hanno già formato oggetto di pronunce tanto di questa Corte che della Corte europea dei diritti dell'uomo»: trattandosi «di questioni di particolare delicatezza, perché coinvolgono, entrambe, valori costituzionali di primario rilievo e vedono i rispettivi modi di concretizzazione reciprocamente implicati»; «al punto che – come è evidente – l'ambito della tutela del diritto all'anonimato della madre non può non condizionare, in concreto, il soddisfacimento della contrapposta aspirazione del figlio alla conoscenza delle proprie origini, e viceversa».
[12] «L'irrevocabilità degli effetti di questa scelta venne», in altre parole, «spiegata secondo una logica di rafforzamento dei corrispondenti obiettivi, escludendo che la decisione per l'anonimato potesse comportare, per la madre, "il rischio di essere, in un imprecisato futuro e su richiesta del figlio mai conosciuto e già adulto, interpellata dall'autorità giudiziaria per decidere se confermare o revocare quella lontana dichiarazione di volontà"»: così, Corte cost., sent. n. 278 cit. (*ibidem*, quarto cpv, secondo per.).

mensione di segretezza[13]. In tale frangente, peraltro, neppure si sarebbe potuto realisticamente discorrere di vero e proprio bilanciamento tra opposti interessi costituzionalmente protetti (che, cronologicamente, si sarebbe avuto, *in limine*, solo quando la donna si fosse risoluta per l'anonimato del parto) giacché la legittimazione di stabilire se mantenere o revocare l'originaria opzione per l'anonimato sarebbe pur sempre stata riconosciuta dall'ordinamento esclusivamente in capo alla madre[14].

Seppure dunque, nella materia *de qua*, paia stagliarsi più degli (e sopra gli) altri il diritto costituzionalmente protetto all'anonimato spettante alla donna, né l'una esigenza – di legittimamente esercitare, cioè, un diritto ad essere dimenticata senza che esso possa in qualche modo subire alcuna interferenza dall'esterno – né l'altra – di scongiurare ogni iniziativa giudiziale volta ad accertare la persistenza della volontà della madre per l'anonimato che possa attentare alla segretezza dell'identità di quest'ultima – sono state alla lunga ritenute realmente concludenti[15]: innanzitutto, perché sussisterebbe il rischio di un endemico ed irreversibile impoverimento del diritto dell'adottato ad accedere al proprio pregresso vissuto non certo meno esiziale di quello cui rimarrebbe esposto il correlativo diritto all'oblio materno; in secondo luogo, giacché sarebbe l'introduzione (astratta) e l'attuazione (in concreto) della possibilità di interpellare la donna invero a condizionare l'effettiva garanzia della *privacy* di quest'ultima[16]. A venire casomai in gioco sarebbe, piuttosto, l'eventuale ripensamento ma-

[13] … «il nucleo fondante della scelta allora adottata» cogliendosi, «così, agevolmente, nella ritenuta corrispondenza biunivoca tra il diritto all'anonimato, in sé e per sé considerato, e la perdurante quanto inderogabile tutela dei profili di riservatezza o, se si vuole, di segreto, che l'esercizio di quel diritto inevitabilmente coinvolge»: così, ancora, Corte cost., sent. n. 278 cit. (*ibidem*, quinto cpv) laddove, in particolare, prosegue ritenendo quest'ultima tutela «un nucleo fondante che – vale la pena puntualizzare – non può che essere riaffermato, proprio alla luce dei valori di primario risalto che esso intende preservare».

[14] «Solo la madre pertanto in questa prospettiva può essere la persona legittimata a decidere se revocare la sua decisione di rimanere anonima in relazione al venir meno di quell'esigenza di protezione che le ha consentito la scelta tutelata dall'ordinamento»: così, Corte cassaz., sez. I civ., sent. 21 luglio 2016, n. 15024 (punto 15 *cons. dir.*) su cui, tra gli altri, G. NALIS, Osservatorio di diritto civile, in *diritto amministrativo,* 28 febbraio 2017; A. GIURLANDA, *op. cit.*; I. LOMBARDINI, *Una questione problematica ancora aperta dopo le recenti pronunce della giurisprudenza: il diritto dell'adottato, non riconosciuto alla nascita, alla conoscenza delle proprie origini e il diritto della madre biologica all'anonimato*, in *diritto,* 6 aprile 2020 e ID., *Il procedimento di "interpello" della madre biologica*, cit., laddove, nello specifico, concorda con quella dottrina «secondo cui, nella specie, il bilanciamento dei diritti fondamentali in gioco appare una categoria inefficace e per certi versi inappropriata [...]»: potendosi, in altre parole «propriamente parlare di ponderazione fra diritti fondamentali» solo «con riferimento al momento della scelta della madre di partorire anonimamente» – «perché in questo momento è in gioco il suo diritto alla vita e quello del figlio» – e non già «dopo la nascita», quando «non è più il diritto alla vita ad essere in gioco e il diritto all'anonimato diventa strumentale a proteggere la scelta compiuta dalle conseguenze sociali e in generale dalle conseguenze negative che verrebbero a ripercuotersi in primo luogo sulla persona della madre» (sul cruciale punto, nondimeno, si tornerà, *infra*, al par. 5, in chiusura del presente contributo).

[15] Il riferimento sarebbe, cioè, a quel «sistema» che – «commisura[ndo] temporalmente lo spazio del "vincolo" all'anonimato a una durata idealmente eccedente quella della vita umana» – «riposa sulla ritenuta esigenza di prevenire turbative nei confronti della madre in relazione all'esercizio di un suo "diritto all'oblio" e, nello stesso tempo, sull'esigenza di salvaguardare *erga omnes* la riservatezza circa l'identità della madre, evidentemente considerata come esposta a rischio ogni volta in cui se ne possa cercare il contatto per verificare se intenda o meno mantenere il proprio anonimato»: così, Corte cost., sent. n. 278 cit. (punto 5 *cons. dir.*, quarto cpv).

[16] Come dire, insomma, che nessuna delle prospettate esigenze è potuta dirsi davvero «dirimente: non la prima, in quanto al pericolo di turbativa della madre corrisponde un contrapposto pericolo per il figlio, depauperato del diritto di conoscere le proprie origini; non la seconda, dal momento che la maggiore o minore ampiez-

terno circa l'assunzione di una genitorialità non più giuridica bensì, quantomeno, naturale: l'originario e risalente diniego della prima non potendo, del resto, astrattamente escludere (*recte*, ben potendo conciliarsi) con la sopravvenuta accettazione della seconda da parte della donna medesima[17].

Passando al differente versante del figlio, non poco utile (e, anzi, oltremodo opportuno) sarebbe preliminarmente distinguere il piano dell'effettiva azionabilità del diritto di apprendere le informazioni relative alla propria precedente storia parentale da quello del relativo parametro di copertura costituzionale.

Prendendo le mosse dal primo profilo, rispetto agli effettivi destinatari del nominato diritto bisogna innanzitutto rilevare come la stessa giurisprudenza costituzionale abbia inizialmente sgombrato il campo da ogni presunta lesione dell'art. 3 Cost. connessa alla diversa regolamentazione del diritto di ricercare le proprie origini a seconda che si tratti dell'adottato figlio di madre che abbia all'epoca optato per l'anonimato ovvero del figlio i cui genitori non si siano mai espressi sul punto (preclusa, cioè, nel primo caso e concessa, invece, nel secondo)[18]: ad essere «caratterizzata dal rapporto conflittuale fra il diritto dell'adottato alla propria identità personale e quello della madre naturale al rispetto della sua volontà di anonimato», difatti, «solo la prima» eventualità sarebbe stata «e non anche la seconda»[19]; con la conseguenza che niente affatto irragionevole si sarebbe dimostrata la persistente differenza di trattamento giuridico dei due richiamati casi[20].

Con riferimento, poi, alla possibile prescrizione del diritto in commento – specie allorquando il richiedente sia già adulto – non difficile sarebbe obiettivamente sostenere, come pure è accaduto, che «l'interesse vitale dell'individuo a ottenere le informazioni necessarie alla scoperta della verità con riguardo ad un aspetto importante della [propria] identità personale, parte integrante del diritto alla vita privata», integri «un diritto soggettivo ed ultra-personale e, pertanto, imprescrittibile»[21]. Conso-

za della tutela della riservatezza resta, in conclusione, affidata alle diverse modalità previste dalle relative discipline, oltre che all'esperienza della loro applicazione»: così, ancora, Corte cost., sent. n. 278 cit. (*ibidem*, quinto cpv).

[17] … «sul piano più generale, una scelta per l'anonimato che comporti una rinuncia irreversibile alla "genitorialità giuridica"» potendo «ragionevolmente non implicare», in altri termini, «anche una definitiva e irreversibile rinuncia alla "genitorialità naturale"»: così, nuovamente, Corte cost., sent. n. 278 cit. (*ibidem*, sesto cpv) laddove, in particolare, si ammette che «ove così fosse, d'altra parte, risulterebbe introdotto nel sistema una sorta di divieto destinato a precludere in radice qualsiasi possibilità di reciproca relazione di fatto tra madre e figlio, con esiti difficilmente compatibili con l'art. 2 Cost.» (*ibidem*, settimo cpv).

[18] … vale a dire, «sotto il profilo dell'irragionevole disparità di trattamento fra l'adottato nato da donna che abbia dichiarato di non voler essere nominata e l'adottato figlio di genitori che non abbiano reso alcuna dichiarazione e abbiano anzi subìto l'adozione»: di latente irragionevolezza era stata, difatti, sospettata «la scelta legislativa di vietare al primo l'accesso alle informazioni sulle proprie origini e consentirla invece al secondo, mentre l'equilibrio dell'adottato e quello dei genitori adottivi [avrebbe potuto] essere esposto nell'ultimo caso ad insidie maggiori che non nel primo, nel quale il genitore biologico a distanza di anni [poteva] avere elaborato la condotta passata», così, Corte cost., sent. n. 425 cit. (punto 6 *cons. dir.*, primo cpv).

[19] Così, Corte cost., sent. n. 425 cit. (*ibidem*, secondo cpv).

[20] Del successivo ripensamento della Corte costituzionale sul pur decisivo punto, si darà comunque conto, *infra*, nel par. 4.

[21] Così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, 25 settembre 2012, par. 54, su cui, *ex plurimis*, D. BUTTURINI, *La pretesa a conoscere le proprie origini come espressione del diritto al rispetto della vita privata*, in *forum costituzionale,* 24 ottobre 2012; R.G. CONTI, *La giurisprudenza civile sull'esecuzione delle deci-*

lidata giurisprudenza di legittimità – ancora molto recentemente[22] – ha, d'altro canto, avuto modo di riconoscere come la piena garanzia del diritto all'identità personale debba necessariamente passare anche attraverso il riconoscimento del diritto «ad uno 'status' filiale corrispondente alla verità biologica»[23]: che l'ordinamento italiano abbia assegnato preminente rilievo a tale ultimo diritto – «in quanto componente essenziale del diritto all'identità personale, in ogni momento della vita di una persona e quindi anche in età adulta»[24] – sarebbe, del resto, ampiamente testimoniato proprio dalla mancata previsione di un vincolo temporale alla legittima attivazione ad opera dell'interessato dell'accertamento della genitorialità da parte del giudice[25].

3. (Segue): nel mosaico costituzionale (tra salute, vita privata e identità individuale)

Venendo al richiamato parametro costituzionale, essenzialmente tre sono state nel tempo le disposizioni che hanno tradizionalmente innervato il diritto in parola, a seconda che a copertura di quest'ultimo si siano invocati l'art. 32, 117, comma 1, ovvero 2 e 3 Cost.

Così – per prendere innanzitutto avvio dalla prima delle previsioni citate – il diritto alla salute psicofisica è stato paritariamente messo in campo sia in relazione al disvelamento del segreto che nell'ipotesi, diametralmente opposta, del suo mantenimento. Per un verso si è, ad esempio, sostenuto che il giudice (in questo caso, EDU) avrebbe dovuto tenere in debita considerazione il non secon-

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sioni della Corte Edu, in *questione giustizia*, 1, 2019, 283 s.; R. TREZZA, *Diritto all'anonimato e diritto a conoscere le proprie origini biologiche*, in *giustizia insieme,* 4 ottobre 2019; I. LOMBARDINI, *Il procedimento di "interpello" della madre biologica*, cit.

[22] Così, Corte cassaz., sez. I civ., sent. 22 settembre 2020, n. 19824 su cui, ad esempio, S. OCCHIPINTI, *Accertamento della maternità, il diritto della madre all'anonimato cessa con la sua morte*, in *altalex,* 2 ottobre 2020; REDAZIONE, *Diritto a conoscere le proprie origini*, in *diritto,* 8 ottobre 2020; L. BONARINI, *Azione giudiziale di accertamento della maternità – parto cd. anonimo. Cass. Civ., sez. I, 22/09/2020, n. 19824*, in *salvis juribus,* 27 novembre 2020, nonché ora, volendo, S. AGOSTA, *Anonimato della madre premorta e riespansione del diritto all'identità personale del figlio (a margine di Cassaz. sent. n. 19824/2020)*, in corso di stampa su *Quad. cost.*, 2021.

[23] … «l'incertezza su tale "status"» potendo «determinare una condizione di disagio ed un "vulnus" allo sviluppo adeguato ed alla formazione della personalità riferibile ad ogni stadio della vita»: con la conseguenza che «la sfera all'interno della quale si colloca il diritto al riconoscimento di uno status filiale corrispondente a verità attiene al nucleo dei diritti inviolabili della persona (art. 2 Cost. e art. 8 CEDU) intesi nella dimensione individuale e relazionale», così, Corte cassaz., sez. I civ., sent. 22 settembre 2020, n. 19824 (punto 2 *cons. dir.*, sesto cpv) richiamando tra le altre, sul punto, Corte cassaz., sez. I civ., sentt. 13 aprile-9 giugno 2015, n. 11887; 29 novembre 2016, n. 24292; 15 febbraio 2017, n. 4020. «Con tali articolate e condivisibili argomentazioni», del resto, manifestamente infondata è stata ritenuta «la questione di legittimità costituzionale dell'art. 270 c.c.» lamentando «che la previsione di imprescrittibilità dell'azione di accertamento giudiziale della paternità o maternità, [avrebbe escluso] qualsiasi possibilità di valutazione da parte del giudice della domanda di dichiarazione giudiziale nei casi in cui l'azione [fosse stata] proposta con notevole ritardo (in quella fattispecie circa quaranta anni), con l'effetto di sacrificare il diritto del presunto padre alla stabilità dei rapporti familiari maturati nel corso del tempo, imponendogli a distanza di molto tempo un accertamento coattivo del rapporto di filiazione che l'interessato avrebbe potuto richiedere prima»: così, nuovamente, Corte cassaz., sez. I civ., sent. 22 settembre 2020, n. 19824 (*ibidem*).

[24] Così, Corte cassaz., sez. I civ., sent. 22 settembre 2020, n. 19824 (*ibidem*, settimo cpv).

[25] … «unitamente a quella che la prova può essere data con ogni mezzo, a norma dell'art. 269 c.c., comma 2»: così, Corte cassaz., *op. et loc. ult. cit.*

dario pericolo che appunto al benessere psico-fisico della persona data in adozione in tenera età (e, al tempo del ricorso, ormai anziana) sarebbe potuto derivare dalla rimozione giudiziaria dell'anonimato intorno alle circostanze della propria nascita[26]. Per un altro, tuttavia, si è invece obiettato che la medesima ricorrente avrebbe «del resto dimostrato un interesse autentico a conoscere l'identità della madre, poiché ha tentato di acquisire una certezza al riguardo»: «un tale comportamento» presupponendo, insomma, «delle sofferenze morali e psichiche, anche se queste non vengono accertate da un punto di vista sanitario»[27] (senza contare il potenziale *vulnus* al medesimo art. 32 cit. che sarebbe potuto discendere al figlio ogni qualvolta si fosse trovato nella materiale impossibilità di accedere a qualsivoglia informazione relativa al corredo genetico parentale[28]).

Con riferimento alla lettura dell'art. 8 CEDU offerta dai giudici di Strasburgo – quando, nella causa *Godelli c. Italia* più volte cit., hanno sanzionato la disciplina italiana per non aver «cercato di stabilire un equilibrio e una proporzionalità tra gli interessi delle parti in causa» e, così facendo, per avere conseguentemente «oltrepassato il margine di discrezionalità che le [era] stato accordato»[29] – oltre che relativamente alle Convenzioni di New York e dell'Aja[30], è stato non di rado invocato poi il parametro di cui all'art. 117, comma 1, Cost. Se, da una parte, si è ritenuto che l'esigenza di conoscere la propria storia personale potesse rientrare tanto nel concetto di vita privata che in quello di vita fami-

[26] «Secondo il Governo» italiano, in particolare, «la Corte» avrebbe dovuto appunto «tenere conto del fatto che la ricorrente, oggi quasi settantenne, è stata adottata all'età di sei anni e che la revoca non consensuale del segreto della sua nascita [avrebbe potuto] rivelarsi difficilissima in questo stadio, considerati i possibili rischi non trascurabili per la sua salute e per la sua famiglia attuale»: così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 58.

[27] Se era, dunque, realistico pensare «che la ricorrente, [al tempo] sessantanovenne, [era] riuscita a costruire la propria personalità anche in assenza di informazioni relative all'identità della madre biologica, si [doveva] ammettere che l'interesse che [poteva] avere un individuo a conoscere la sua ascendenza non [veniva] meno con l'età, anzi [avveniva] il contrario»: così, ancora, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 69, sul punto richiamando – tra i suoi precedenti in materia – Corte europea dei diritti dell'uomo, sez. terza, *Jäggi c. Svizzera*, 13 luglio 2006, spec. par. 40, su cui, *ex multis*, C. CAMPIGLIO, *Con la morte, l'uomo perde il diritto al rispetto della vita privata* e S. TONOLO, *Identità personale, maternità surrogata e superiore interesse del minore nella più recente giurisprudenza della Corte europea dei diritti dell'uomo*, entrambe in *Dir. umani e dir. internaz.*, rispettivamente, 2, 2007, 394 ss. e 1, 2015, 202 ss.; L. POLI, *Il diritto a conoscere le proprie origini e le tecniche di fecondazione assistita: profili di diritto internazionale*, in *GenIUS*, 1, 2016, 43 ss.

[28] … «in ragione dell'impossibilità», in altre parole, «per il figlio, di ottenere dati relativi all'anamnesi familiare, anche in relazione al rischio genetico»: «in quanto l'impedimento alla conoscenza dei dati inerenti alla madre naturale priverebbe l'adottato di qualsiasi possibilità di ottenere una anamnesi familiare, essenziale per interventi di profilassi o di accertamenti diagnostici, essendo già egli privo di notizie circa la storia sanitaria del ramo paterno del proprio albero genealogico. Ciò, peraltro, in costanza della prassi, diffusa negli ospedali italiani, di omettere la stessa ordinaria raccolta dei dati anamnestici non identificativi della madre» [così, Corte cost., sent. n. 278 cit. (rispettivamente, punto 1 *cons. dir.*, secondo cpv e punto 1 *rit. fatto*, ottavo cpv)].

[29] Così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 71 espressamente richiamata da Corte cost., sent. n. 278 cit. (punto 1 *cons. dir.*, secondo cpv).

[30] … per «violazione», rispettivamente, «degli artt. 7 e 8 della Convenzione di New York sui diritti del fanciullo del 20 ottobre 1989, resa esecutiva con la L. n. 176 del 1991, laddove si impone il rispetto dei diritti del minore ivi compresi quelli volti a preservare la sua identità, il suo nome e le sue relazioni familiari» («per l'adottato l'identità» consistendo «proprio nel ricercare le proprie origini, le proprie radici e conoscere le informazioni relative alla famiglia biologica») nonché «dell'art. 30, della Convenzione dell'Aja 29 maggio 1993, resa esecutiva con la L. n. 476 del 1998»: così, Corte cassaz., sez. I civ., sent. 29 maggio 2017-20 marzo 2018, n. 6963 (punto 4.1 *cons. dir.*).

liare (profili entrambi protetti, com'è noto, dall'art. 8 cit.)[31], dall'altra, tuttavia, la Corte EDU ha chiaramente dimostrato invece di voler limitare alla sola vita privata il richiamo alla predetta disposizione. Anche attraverso il rinvio a taluni celebri precedenti in materia[32], difatti, non troppo difficile si è dimostrato in quell'occasione rilevare come ad essere stata avanzata fu solo la richiesta di accedere all'identità dei propri ascendenti biologici (quale componente importante per la ricostruzione della propria identità personale) e non pure quella di verificare la propria condizione adottiva (invece riconducibile, appunto, alla nozione di vita familiare)[33].

Interpellati, insomma, sul punto i giudici convenzionali non potevano che ricordare «che "l'articolo 8 tutela un diritto all'identità e allo sviluppo personale e quello di allacciare e approfondire relazioni con i propri simili e il mondo esterno"»: «a tale sviluppo», cioè, contribuendo «la scoperta dei dettagli relativi alla propria identità di essere umano e l'interesse vitale, tutelato dalla Convenzione, a ottenere delle informazioni necessarie alla scoperta della verità riguardante un aspetto importante dell'identità personale, ad esempio l'identità dei propri genitori»[34]. Da quest'ultimo punto di vista, la

[31] … «la ricorrente» avendo in particolare sostenuto «che la sua richiesta di ottenere informazioni su aspetti eminentemente personali della sua storia e della sua infanzia [rientrasse] nel campo di applicazione dell'articolo 8 della Convenzione» giacché «la ricerca della sua identità [faceva] parte integrante della sua "vita privata" ma anche della sua "vita famigliare"»: così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 43.

[32] Part. casi *Mikulić c. Croazia*, 7 febbraio 2002, par. 53, su cui, tra i tanti, C. CAMPIGLIO, *Il divieto di fecondazione eterologa all'esame della Corte europea dei diritti umani*, in *Dir. umani e dir. internaz.*, 3, 2010, spec. 4, D. BUTTURINI, *op. cit.*, part. 3, A. CIERVO, *Il diritto all'anonimato della madre biologica ovvero quando Strasburgo anticipa Roma*, in https://diritti-cedu.unipg.it/, 15 febbraio 2014; par. 2, e *Odièvre c. Francia*, 13 febbraio 2003, par. 29 su cui, *ex plurimis*, J. LONG, *Ammissibilità del parto anonimo e accesso alle informazioni sulle proprie origini: il caso Odièvre c. Francia (introduzione a Corte europea per i diritti dell'uomo, sentenza 13 febbraio 2003, Odièvre c. Francia)*, in *Minori e giustizia*, 3, 2003, 172 ss.; A. RENDA, *La sentenza O c. Francia della Corte Europea dei diritti dell'uomo: un passo indietro rispetto all'interesse a conoscere le proprie origini biologiche*, in *Familia*, 4, 2004, 1121 ss.; S. FAVALLI, *op. cit.*

[33] «Nella fattispecie», insomma, «la ricorrente non chiede[va] di rimettere in questione l'esistenza della sua filiazione adottiva, ma di conoscere le circostanze della sua nascita e del suo abbandono, che comprend[evano] la conoscenza dell'identità dei suoi genitori biologici»: con la conseguenza che «la Corte» non fosse «chiamata a determinare se la procedura che riguarda[va] il legame di filiazione tra la ricorrente e la madre rientr[asse] nella "vita famigliare" ai sensi dell'articolo 8, poiché in ogni caso il diritto di conoscere la propria ascendenza rientra[va] nel campo di applicazione della nozione di "vita privata" che comprende[va] aspetti importanti dell'identità personale di cui fa[ceva] parte l'identità dei genitori» (così, Corte europea dei diritti dell'uomo, *ibidem*, par. 45). Sul precipuo punto, peraltro, vale la pena ricordare che «il Governo [aveva sostenuto] che non [esistesse] tra la ricorrente e la madre biologica alcuna vita famigliare ai sensi dell'articolo 8 della Convenzione, in quanto la prima non [aveva] mai visto la madre, poiché quest'ultima non [aveva] mai voluto conoscerla e considerarla come sua figlia» («in effetti, essa» avendo «espressamente manifestato la propria volontà di abbandonarla» ed «accettato che la figlia venisse adottata»): «garantendo il diritto al rispetto della vita famigliare, l'articolo 8 presuppone[va]», d'altro canto, «l'esistenza di una famiglia (*Marckx c. Belgio*, sentenza del 13 giugno 1979, serie A n. 31)»; «se la giurisprudenza non [esigeva] che vi [fosse] convivenza tra i vari membri della "famiglia", [sarebbero dovuti] sussistere quantomeno dei rapporti personali stretti tra di essi» i quali avrebbero dimostrato «una relazione affettiva tra due esseri e la loro volontà di intrattenere tale relazione sarebbe [stata] fondamentale per gli organi della Convenzione» («questi ultimi» ritenendo «anche che il solo legame biologico [fosse] insufficiente, in assenza di legami personali stretti tra gli interessati, per costituire una vita famigliare ai sensi dell'articolo 8») (così, *ibidem*, par. 44).

[34] Con la conseguenza che «la nascita, e in particolare le circostanze di quest'ultima, rientra nella vita privata del bambino, e poi dell'adulto, sancita dall'articolo 8 della Convenzione che trova così applicazione nel caso di

Corte europea non ha peraltro perso l'occasione di rimarcare come tra i doveri discendenti sul singolo ordinamento nazionale dall'art. 8 cit. nemmeno potesse dirsi esistente una netta demarcazione tra quelli aventi natura negativa ovvero meramente positiva: al contrario, non difficilmente questi ultimi – si pensi, appunto, alla necessità di adottare ogni provvedimento volto ad assicurare l'effettività della tutela della vita privata – si sarebbero potuti sovrapporre al mero obbligo di astensione statale da ogni abusiva interferenza pubblica nella sfera privata della persona[35].

Sebbene dunque abbia *formalmente* dichiarato l'assorbimento di ogni altra eccezione relativa all'ulteriore parametro di cui all'art. 117, comma 1, cit.[36], pochi dubbi possono in effetti nutrirsi sul fatto che la stessa giurisprudenza costituzionale sembra aver finito per *materialmente* recepire quanto già in effetti osservato a livello convenzionale: specie quando si è rilevata la diversità di esiti tra la normativa francese e quella italiana rispetto alla comune esigenza di aprire uno – sia pur minimo – spiraglio al diritto dell'adottato di accedere al proprio pregresso vissuto[37].

Passando adesso ai più volte nominati artt. 2 e 3 Cost., è innanzitutto ovvio come – sul piano del metodo[38] – non possa *mai* escludersi (e, anzi, debba *sempre* imporsi) un'interpretazione della disciplina

specie»: così – invocando pure il precedente di cui a *Mikulić c. Croazia*, cit., parr. 54 e 64 – Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 46, richiamata pure da Corte cassaz., sez. I civ., sent. 21 luglio 2016, n. 15024 (punto 9 *cons. dir.*) laddove, in particolare, ricorda come «la Corte europea dei diritti dell'uomo [...] ha dato una interpretazione dell'art. 8 della Convenzione E.D.U., che riconduce il diritto alla conoscenza delle proprie origini nell'ambito di applicazione della nozione di vita privata e specificamente nella sfera di protezione dell'identità personale», «in questa prospettiva» affermando «che l'art. 8 protegge il diritto all'identità e alla realizzazione personale e quello di intessere e sviluppare relazioni con i propri simili e il mondo esterno».

[35] «Se l'articolo 8 tende fondamentalmente a difendere l'individuo da ingerenze arbitrarie dei pubblici poteri, esso», tuttavia, «non si limita ad ordinare allo Stato di astenersi da ingerenze di questo tipo: a questo impegno piuttosto negativo possono aggiungersi obblighi positivi inerenti a un rispetto effettivo della vita privata. Essi possono implicare l'adozione di misure volte al rispetto della vita privata fino alle relazioni degli individui tra loro (*X e Y c. Paesi Bassi*, sentenza del 26 marzo 1985, § 23, serie A n. 91)»; d'altra parte, «la linea di separazione tra gli obblighi positivi e negativi dello Stato a titolo dell'articolo 8 non si presta ad essere definita con precisione; i principi applicabili sono comunque assimilabili. In particolare, in entrambi i casi, si deve avere riguardo al giusto equilibrio da mantenere tra gli interessi concorrenti; parimenti, in entrambe le ipotesi lo Stato gode di un certo margine di discrezionalità (*Mikulić* sopra citata, § 58)»: così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 60, sul punto espressamente richiamata anche da Corte cassaz., sez. I civ., sent. 22 settembre 2020, n. 19824 (punto 2 *cons. dir.*, sedicesimo cpv) quando evidenzia come «l'art. 8 CEDU, nella lettura datane dalla Corte EDU (Corte EDU, 22/09/2012, Godelli c. Italia, Corte EDU, 13/02/2003, Odievre c. Francia), tende essenzialmente a premunire l'individuo contro ingerenze arbitrarie dei poteri pubblici, non contentandosi di ordinare allo Stato di astenersi da simili ingerenze, ma aggiungendovi obblighi positivi inerenti ad un rispetto effettivo della vita privata; tra questi non può non rientrare il diritto a proporre le azioni che lo stesso ordinamento nazionale offre per il riconoscimento dello status di figlio naturale di una persona».

[36] Così, Corte cost., sent. n. 278 cit. (punto 6 *cons. dir.*, settimo cpv).

[37] «Ciò, d'altra parte, risulta sulla base degli stessi rilievi, in sostanza, formulati dalla Corte EDU nella richiamata "sentenza Godelli"» allorché «si è stigmatizzato che la normativa italiana non darebbe "alcuna possibilità al figlio adottivo e non riconosciuto alla nascita di chiedere l'accesso ad informazioni non identificative sulle sue origini o la reversibilità del segreto", a differenza di quanto, invece, previsto nel sistema francese, scrutinato, in parte qua, nella sentenza 13 febbraio 2003, nel 'caso Odièvre'»: così, Corte cost., sent. n. 278 cit. (*ibidem*, rispettivamente, secondo e terzo cpv).

[38] Così per esempio, ad esempio, A. RAUTI, *La "cerchia dei custodi" delle "Carte" nelle sentenze costituzionali nn. 348-349 del 2007: considerazioni problematiche*, in C. SALAZAR, A. SPADARO (a cura di), *Riflessioni sulle sentenze*

interna dell'anonimato che possa doppiamente mostrarsi conforme a Costituzione (part. artt. 2, 24 e 30) e a CEDU (*ex* art. 117 cit.)[39]. Se la conoscenza delle circostanze intorno alla propria venuta al mondo ed alla successiva separazione dalla madre – non meno delle relazioni affettive instaurate – contribuisce alla compiuta realizzazione della propria storia parentale è, difatti, certo che il diritto alla ricerca delle origini non possa che ambientarsi e più profondamente attecchire entro il perimetro delineato dalle previsioni costituzionali così come, pure, da quelle EDU a protezione della vita privata e familiare[40].

4. (Segue): nella dimensione dinamica

Passando al connesso piano del merito, è stato evidenziato come il *vulnus* agli artt. 2 e 3 Cost. – col discendente obbligo costituzionale di rimozione – venga invero inflitto dall'imposizione legislativa di un anonimato materno assoluto ed incondizionato[41]: il quale finirebbe così non solo per porsi diametralmente in contrasto col «diritto di ricerca delle proprie origini e dunque del diritto all'identità personale dell'adottato» ma, pure, per irragionevolmente discriminare nel trattamento giuridico «fra l'adottato nato da donna che abbia dichiarato di non voler essere nominata e l'adottato figlio di genitori che non abbiano reso alcuna dichiarazione e abbiano anzi subìto l'adozione»[42].

Con tali premesse, se si volesse adesso riguardare ai valori costituzionali in campo – come ci si era d'altro canto ripromesso di fare *supra* – in senso dinamico, indubbio appare il largo spazio di discrezionalità spettante agli ordinamenti nazionali ogni qualvolta siano chiamati a «scegliere i mezzi che ritengono più idonei ad assicurare in modo equo la conciliazione tra la protezione della madre e la richiesta legittima» di conoscere la propria storia personale «nel rispetto dell'interesse generale»[43] (specie quando, proprio intorno al contrasto tra le contrapposte esigenze di cui sono portatori due soggetti privati, non possa dirsi maturato un sufficiente margine di condivisione tra gli Stati europei[44]). Nel precipuo caso dell'art. 8 CEDU non pochi sono, del resto, i fattori condizionanti il più o

348-349/2007 della Corte costituzionale, Milano, 2009, spec. 310; G. ROLLA, *Il processo di ibridazione dei sistemi accentrati di giustizia costituzionale. Note di diritto comparato*, in M. CARBONELL SÁNCHEZ, H. FIX ZAMUDIO, L. RAÚL GONZÁLEA PÉREZ, D.VALADÉS RÍOS (a cura di), *Estado constitucional, derechos humanos, justicia y vida universitariaEstudios en homenaje a Jorge Carpizo*, Messico, 2015, part. 529; P. COSTANZO, L. MEZZETTI, A. RUGGERI, *Lineamenti di diritto costituzionale dell'Unione europea*, Torino, 2019, spec. 287 ss.

[39] Così, Corte cassaz., sez. I civ., sent. 22 settembre 2020, n. 19824 (punto 2 *cons. dir.*, sedicesimo cpv).

[40] Se «la cornice costituzionale e convenzionale del diritto a conoscere le proprie origini, quale declinazione di primario rilievo del diritto all'identità personale, è costituita dagli articoli 2 e 3 Cost., e 8 della Corte Edu» non vi è, in altri termini, dubbio che «lo sviluppo della personalità individuale e l'armonica conduzione della propria vita privata e familiare richiedono la costruzione di una propria identità individuale fondata, oltre che su un contesto parentale affettivo-educativo riconoscibile, anche su informazioni relative alla propria nascita idonee a svelarne il segreto unitamente alle ragioni dell'abbandono»: così, Corte cassaz., sez. I civ., sent. 9 novembre 2016, n. 22838 (punto 4.1 *cons. dir.*, primo cpv).

[41] Così, Corte cost., sent. n. 278 cit. (punto 6 *cons. dir.*, sesto cpv).

[42] Così, nuovamente, Corte cost., sent. n. 278 cit. (punto 1 *cons. dir.*, secondo cpv).

[43] Così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 67.

[44] A ritenere «che, quando due interessi privati entrano in conflitto, lo Stato dispone di un certo margine di discrezionalità» è stato, in particolare, il Governo italiano nel momento in cui ha sostenuto che quest'ultimo «è del resto rafforzato, nella presente causa, in quanto non esiste sulla questione dell'accesso del figlio ad infor-

meno esteso potere discrezionale statale (tra i quali non può che spiccare la specifica prospettiva della vita privata venuta, di volta in volta, in gioco nel caso concreto)[45].

Quanto appena esposto, tuttavia, non preclude naturalmente all'autorità giurisdizionale – e non potrebbe, d'altronde, essere altrimenti – un controllo successivo sulle misure legislative precipuamente adottate: il quale ultimo, se nel *metodo* inevitabilmente si differenzia a seconda che ad intervenire siano stati i giudici EDU ovvero quelli costituzionali italiani, è nel *merito* che piuttosto esibisce interessanti sovrapposizioni argomentative tra giurisprudenza convenzionale e domestica. Così per esempio, sul piano metodico, l'anonimato materno dovrebbe in effetti imporre alla Corte di Strasburgo di non sovrapporsi all'autorità interna nell'individuare quale sia lo strumento più adeguato alla disciplina di siffatta questione, dovendo quest'ultima necessariamente attenersi alla vicenda concretamente venuta al suo esame, senza estendere pure il proprio sindacato alla regolazione statale astrattamente e nel suo complesso considerata[46].

Ma se, come si anticipava, di vero e proprio accavallamento può discorrersi esso pare emergere sotto il diverso profilo del merito, a partire dalla funzione dello stesso *scrutiny* richiesto. Il quale sindacato – proprio in virtù del fatto che il concetto di vita privata non possa fare a meno del diritto all'identità personale e quest'ultimo, a sua volta, di quello all'accesso al proprio pregresso vissuto, in una sorta di reciproco gioco a scatole cinesi di rimandi – dovrà poi dimostrarsi, per così dire, più *strict* del consueto nel soppesare ogni esigenza costituzionale sul tappeto[47] e, in ultima istanza, nel determinare se tra il diritto materno al parto anonimo e quello del figlio ad apprendere le informazioni relative alla propria precedente storia parentale sia stata individuata la migliore condizione di bilanciamento alle condizioni storiche e di contesto date[48]: «il diritto all'identità, come condizione essenziale del diritto

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mazioni sulle proprie origini alcun consenso a livello europeo»: così, ancora, Corte europea dei diritti dell'uomo, *ibidem*, par. 59.

[45] Gli stessi giudici di Strasburgo, difatti, ricordano «che la scelta delle misure idonee a garantire il rispetto dell'articolo 8 della Convenzione nei rapporti interpersonali rientra in linea di principio nel margine di discrezionalità degli Stati contraenti», esistendo «a tale proposito vari modi di assicurare il rispetto della vita privata e la natura dell'obbligo dello Stato [dipendendo] dall'aspetto della vita privata che viene messo in discussione (Odièvre, sopra citata, § 46)»: «l'ampiezza di tale margine di discrezionalità dello Stato dipende non solo dal o dai diritti interessati ma anche, per ciascun diritto, dalla natura stessa di ciò che viene messo in causa», in tal senso, Corte europea dei diritti dell'uomo, *ibidem*, par. 65 (secondo e terzo per.).

[46] «In una causa originata da un ricorso individuale», insomma «la Corte non ha il compito di controllare in astratto una legislazione o una prassi contestate, ma deve limitarsi il più possibile, senza tralasciare il contesto generale, ad esaminare le questioni sollevate dal caso concreto di cui si trova investita»: «essa non deve quindi sostituire la sua valutazione a quella delle autorità nazionali competenti per stabilire quale sia il mezzo migliore per regolamentare le questioni (S. H. e altri c. Austria [GC], n. 57813/00, § 92, CEDU 2011) che pone il parto anonimo»; non spettando, vale a dire, ad essa «controllare la necessità del divieto assoluto, giudicata costituzionale dal legislatore italiano, dal momento che questa misura non è arbitraria e che il bilanciamento tiene ragionevolmente conto di tutti i diritti in gioco» [così, *dissenting opinion* del giudice A. Sajó (*ibidem*), cit.].

[47] Così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., *ibidem* (quarto per.)

[48] «In una situazione in cui sono in conflitto i diritti, sanciti dalla Convenzione, di due titolari di diritti, il ruolo della Corte è», difatti, «quello di vigilare affinché nella causa venga mantenuto un giusto equilibrio»: «questo presuppone che alle autorità nazionali venga lasciato un adeguato margine di discrezionalità ai fini di un bilanciamento, avendo la Corte un ruolo di vigilanza. "Se il bilanciamento da parte delle autorità nazionali è operato nel rispetto dei criteri stabiliti dalla giurisprudenza della Corte, occorrono motivi seri perché quest'ultima sostituisca il suo parere a quello dei giudici interni" (Von Hannover c. Germania (n. 2) [GC], nn. 40660/08 e 60641/08, § 107, CEDU 2012)» (così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par.

all'autonomia (*Pretty c. Regno Unito*) e allo sviluppo della persona (*Bensaid c. Regno Unito*)», d'altro canto, «fa parte del nocciolo duro del diritto al rispetto della vita privata e pertanto un esame tanto più rigoroso si impone per bilanciare effettivamente gli interessi in gioco»[49].

Alla luce di tale rigoroso scrutinio è perciò ovvio che in principio ritenuta non in conflitto con l'art. 2 Cost. – nella misura in cui avrebbe individuato un corretto equilibrio tra tutte le istanze espresse in giudizio[50] – quella (temporalmente incondizionata) protezione inizialmente accordata al diritto materno al parto anonimo[51] finisca inevitabilmente per diventarlo tanto per la giurisprudenza EDU (2012) che per quella costituzionale di poco successiva (2013), in entrambi i frangenti essendo proprio la medesima rigidità del bilanciamento così realizzato ritenuta del tutto sproporzionata e, dunque, eccessiva.

Che vi sia intanto stata un'intollerabile violazione dell'art. 8 CEDU ai giudici di Strasburgo è apparso innanzitutto chiaro dal momento che era appunto risultata un'illimitata prevalenza dell'esigenza materna a conservare l'anonimato circa la propria identità rispetto al concorrente interesse del figlio alla ricerca delle proprie origini senza che il legislatore italiano si fosse neppure sforzato di individuare quel *minimum* di congegni normativi, per così dire, di riequilibrio confezionati invece, in materia, dalla corrispondente disciplina transalpina[52]. «Se la madre biologica [avesse] deciso di mantenere l'anonimato, la normativa italiana non [avrebbe dato] alcuna possibilità al figlio adottivo e non riconosciuto alla nascita di chiedere l'accesso ad informazioni non identificative sulle sue origini o la reversibilità del segreto»: con l'ineludibile conseguenza che – «non [avendo] cercato di stabilire un

66). «La scelta dei mezzi più adatti per assicurare equamente la conciliazione dell'istanza di protezione della madre, che si trova in una condizione di difficoltà tale da non consentirle di assumere il ruolo genitoriale, con la domanda legittima del figlio ad avere accesso alle informazioni sulle sue origini spetta», in altre parole, «agli Stati aderenti alla Convenzione»: «tuttavia, la Corte è nelle condizioni di esercitare un sindacato circa la scelta e l'effettivo esercizio di tali mezzi di composizione del conflitto e, in particolare, sulla ricerca e la realizzazione di un equilibrio fra i concorrenti interessi e diritti in gioco»: così, Corte cassaz., sez. I civ., sent. 21 luglio 2016, n. 15024 (punto 11 *cons. dir.*)

[49] Così, nuovamente, Corte cassaz., sez. I civ., sent. 21 luglio 2016, n. 15024 (punto 13 *cons. dir.*, ottavo per.).

[50] Così, Corte cost., sent. n. 425 cit. (*ibidem*, quinto cpv).

[51] A motivo del fatto che, come visto nelle pagine che precedono, «la scelta della gestante in difficoltà che la legge vuole favorire – per proteggere tanto lei quanto il nascituro – sarebbe resa oltremodo difficile se la decisione di partorire in una struttura medica adeguata, rimanendo anonima, potesse comportare per la donna, in base alla stessa norma, il rischio di essere, in un imprecisato futuro e su richiesta di un figlio mai conosciuto e già adulto, interpellata dall'autorità giudiziaria per decidere se confermare o revocare quella lontana dichiarazione di volontà»: così, Corte cost., sent. n. 425 cit. (punto 4 *cons. dir.*, quarto cpv).

[52] Si è difatti osservato «che, a differenza del sistema francese esaminato nella sentenza Odièvre, la normativa italiana non [tentava] di mantenere alcun equilibrio tra i diritti e gli interessi concorrenti in causa. In assenza di meccanismi destinati a bilanciare il diritto della ricorrente a conoscere le proprie origini con i diritti e gli interessi della madre a mantenere l'anonimato, [veniva] inevitabilmente data una preferenza incondizionata a questi ultimi»: così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 70, sempre riportandosi al suo precedente *decisum* nel caso *Odièvre* cit. quando ha considerato «che la nuova legge del 22 gennaio 2002 aumenta[va] la possibilità di revocare il segreto dell'identità e agevola[va] la ricerca delle origini biologiche grazie alla creazione di un Consiglio nazionale per l'accesso alle origini personali» [«di immediata applicazione, essa permette[va] ormai alle persone interessate di chiedere la reversibilità del segreto dell'identità della madre, a condizione che quest'ultima vi acconsent[isse] (§ 49), nonché di avere accesso a informazioni non identificative»].

equilibrio e una proporzionalità tra gli interessi delle parti in causa» – l'Italia aveva perciò finito per oltrepassare «il margine di discrezionalità che le [era] stato accordato»[53].

5. Centralità del veto materno e residue ambiguità di un bilanciamento (mascherato)

Di una vera e propria «cristallizzazione», ovvero «immobilizzazione», nei precipui modi di applicazione del diritto all'anonimato della madre prefigurata dall'ordinamento[54] ha invece discorso la giurisprudenza costituzionale allorché ha censurato la regolamentazione interna per l'assolutezza delle sue previsioni[55]. Da quando veniva esercitato dalla madre, il diritto al parto anonimo finiva difatti, per un verso, per tradursi in un irreversibile ostacolo giuridico per il figlio che desiderasse conoscere la propria storia personale ma, per un altro, per ritorcersi in una sorta di vera e propria "espropriazione" in danno della donna medesima: la quale, da quel momento in avanti, si sarebbe suo malgrado ritrovata spogliata di qualsivoglia opzione alternativa da quello stesso ordinamento che, in tesi, mirava invece a tutelarla[56].

All'esito del non poco travagliato tragitto giurisprudenziale del quale si è cercato, seppur succintamente, di dar conto nelle pagine che precedono, un più o meno ampio varco tra le strette maglie di cui è normativamente intessuto il diritto all'anonimato materno sembra infine essere stato dal diritto vivente aperto, laddove è stato ad esempio ammesso il legittimo esperimento dell'azione volta giudizialmente ad accertare il legame con la donna ogniqualvolta quest'ultima avesse, nei fatti, sconfessato l'originaria decisione di abdicare alla propria maternità giuridica: al di fuori di tali frangenti restando, nondimeno, intesa la massima e duratura protezione del segreto materno intorno al parto[57].

Alla luce di tutto quanto sopradetto, insomma, ancora di recente pare pienamente confermarsi l'indiscussa prevalenza da accordare al diritto al parto anonimo – per l'intero arco dell'esistenza della donna – ogni volta che dovesse entrare nel bilanciamento costituzionale col concorrente diritto del figlio a vedersi riconosciuto il proprio personale vissuto: ciò evidentemente ancora sulla scorta del futuro rischio di disvelamento del segreto che la prima potrebbe mettere in conto in un momento assai

[53] Così, Corte europea dei diritti dell'uomo, sez. seconda, *Godelli c. Italia*, cit., par. 71.

[54] Così, Corte cost., sent. n. 278 cit. (punto 5 *cons. dir.*, secondo cpv).

[55] Così, Corte cost., sent. n. 278 cit. (punto 6 *cons. dir.*, primo cpv).

[56] «Una volta intervenuta la scelta per l'anonimato, infatti, la relativa manifestazione di volontà assume[va] connotati di irreversibilità destinati, sostanzialmente, ad "espropriare" la persona titolare del diritto da qualsiasi ulteriore opzione; trasformandosi, in definitiva, quel diritto in una sorta di vincolo obbligatorio, che fini[va] per avere un'efficacia espansiva esterna al suo stesso titolare e, dunque, per proiettare l'impedimento alla eventuale relativa rimozione proprio sul figlio, alla posizione del quale si [era] inteso, ab origine, collegare il vincolo del segreto su chi lo [avesse] generato»: così, Corte cost., sent. n. 278 cit. (punto 5 *cons. dir.*, secondo cpv).

[57] … «tale regola» potendo insomma essere, «al limite, derogata (consentendo quindi l'esercizio dell'accertamento giudiziale della maternità) solo ove fosse stata proprio la madre [...] con la propria inequivocabile condotta, ad aver manifestato la volontà di revocare nei fatti la scelta, a suo tempo presa, di rinuncia alla genitorialità giuridica, accogliendo nella propria casa il bambino come un figlio»: «tuttavia, al di fuori del caso limite sopra enunciato, la tutela del diritto all'anonimato della madre, per tutta la durata della vita della stessa, deve essere, come detto, massima», così, Corte cassaz., sez. I civ., sent. 22 settembre 2020, n. 19824 (punto 2 *cons. dir.*, rispettivamente, decimo ed undicesimo cpv).

delicato (come, appunto, la nascita) e che potrebbe piuttosto spingerla ad irreversibili decisioni per la salute e la vita di sé stessa e del nascituro che porta in grembo[58].

A margine di queste poche sparse notazioni, ad ogni modo, non può certo sfuggire quel complesso di luci ed ombre che ha inevitabilmente accompagnato (e tutt'ora segue da assai vicino…) il tentativo italiano di flessibilizzazione dell'originaria preclusione di apprendere le informazioni relative alla propria precedente storia parentale per via giurisprudenziale e che finisce tutto sommato per accomunarlo a quello della vicina esperienza transalpina (dove tale temperamento si è piuttosto avuto, come detto, per via legislativa). Così ad esempio, anche alla complessa vicenda nostrana, possono non troppo difficilmente estendersi quelle perplessità già in precedenza avanzate dalla giurisprudenza EDU a proposito del caso francese tanto nel *metodo* che nel *merito* osservato[59].

Se – favorendo un eventuale ripensamento materno dell'iniziale scelta per l'anonimato – il diritto vivente italiano ha indiscutibilmente determinato, per un verso, una timida apertura in favore del diritto dell'adottato alla ricerca delle proprie origini (come parallelamente accaduto, del resto, in Francia con la più volte cit. l. n. 93/2002, *relative à l'accès aux origines des personnes adoptées et pupilles de l'Etat*[60]), per un altro, tuttavia, già sul piano del merito non vi è dubbio come anche per l'Italia possa valere la considerazione che «tale reversibilità [sia] in ultima istanza affidata e condizionata dall'accordo» della madre, quest'ultima «[essendo] solo invitata e non [avendo] l'obbligo di rilasciare delle indicazioni identificative»[61]. Mentre sul piano giuridico diventa così inevitabile che tale unilaterale decisione materna possa essere opposta dal diretto interessato[62] (ovvero rimossa da un organo terzo[63]), su quello più strettamente psicologico pochi dubbi possono invero nutrirsi sul fatto che la

[58] «Nel bilanciamento dei valori di rango costituzionale che si impone all'interprete, al cospetto del diritto al riconoscimento dello *status* di filiazione, quello della madre a mantenere l'anonimato al momento del parto si pon[e] comunque in posizione preminente»: «quest'ultimo diritto, infatti, [...] è finalizzato a tutelare i beni supremi della salute e della vita, oltre che del nascituro, della madre, la quale potrebbe essere indotta a scelte di natura diversa, fonte di possibile forte rischio per entrambi, ove, nel momento di estrema fragilità che caratterizza il parto, la donna che opta per l'anonimato avesse solo il dubbio di poter essere esposta, in seguito, ad un'azione di accertamento giudiziale della maternità»: così, ancora recentemente, Corte cassaz., sez. I civ., sent. 22 settembre 2020, n. 19824 (*ibidem*, rispettivamente, ottavo e nono cpv).

[59] «La citata sentenza della Corte europea *Odièvre c. Francia*, di cui la sentenza *Godelli c. Italia* è la coerente riaffermazione» già rappresentava, d'altronde, «un precedente sofferto perché è stato pronunciato all'esito della ricerca di un difficile equilibrio fra tradizioni giuridiche e posizioni di principio molto diverse come è eloquentemente rappresentato nella opinione dissenziente dei giudici Wildhaber, Bratza, Bonello, Loucaides, Cabral Barreto, Tulkens e Pellonpää»: così, Corte cassaz., sez. I civ., sent. 21 luglio 2016, n. 15024 (punto 13 *cons. dir.*, primo per.).

[60] La disciplina francese sottoposta allo scrutinio EDU «riconosce chiaramente la necessità di trovare un riequilibrio dei diritti in conflitto»: «essa, pur non mettendo in discussione l'istituto dell'*accouchement sous x*, segna certamente un passo in avanti in materia di accesso alla conoscenza delle proprie origini in quanto consente di sollecitare la reversibilità del segreto sull'identità della madre» (così, Corte cassaz., *op. et loc. ult. cit.*, nono e decimo per.).

[61] … così come nell'ordinamento francese dove, d'altra parte, costei «può sempre opporsi a che la sua identità sia svelata anche dopo la sua morte (articolo L. 147-6 del '*code de l'action sociale et des familles*' introdotto dall'art. 1 della l. 22 gennaio 2002)»: così, Corte cassaz., *op. et loc. ult. cit.*, undicesimo e dodicesimo per.

[62] … «il rifiuto della madre» imponendosi, difatti, «al figlio che non ha alcun mezzo giuridico per contrastare la sua volontà unilaterale»: così, Corte cassaz., *op. et loc. ult. cit.*, quarto per.

[63] Come, appunto, in Francia dove non è stato «previsto che il Consiglio Nazionale che ha istituito (né alcun altro organo indipendente) possa prendere una decisione finale sulla rimozione del segreto, in considerazione

donna finisca per vedersi riconosciuto «il diritto puramente discrezionale di mettere al mondo un bambino ponendolo in una condizione di sofferenza e condannandolo per tutta la vita all'ignoranza sulle sue origini»[64].

Al di là di tali considerazioni, nondimeno, se c'è un profilo che, più degli altri, ha attirato gli strali della dottrina più perplessa esso è senz'altro stato quello metodico. Laddove si è criticamente osservato cioè – nella richiamata giurisprudenza *Odièvre* (ma le riflessioni sono pianamente estendibili, come si è visto, anche all'odierna esperienza italiana) – come impropriamente potrebbe in tale frangente discorrersi di ragionevole ponderazione tra tutte le istanze costituzionali in gioco giacché la persistente validità della disciplina sull'anonimato materno continuerebbe piuttosto a mantenere una non rimossa diseguaglianza di fondo tra i trattamenti di esse[65]. Nel momento in cui si ammetta in conclusione – a prescindere dalle motivazioni che la sorreggono – che la scelta materna possa costituire per l'adottato uno sbarramento invalicabile sulla strada della conoscenza della propria storia personale[66], è perciò inevitabile il rischio che continui a perpetrarsi (con legge, in Francia, così come, specularmente, con la giurisprudenza, in Italia) un palese disequilibrio tra tutte le esigenze coinvolte: «il diritto di veto puro e semplice riconosciuto alla madre [comportando] che i diritti del minore riconosciuti nel sistema generale della convenzione (sentenze *Johansen c. Norvège*, *Kuzner c. Germania*), [siano] interamente negati e dimenticati»[67].

degli interessi in conflitto, nell'ipotesi in cui la madre permanga nella sua posizione di rifiuto che comporta la definitiva privazione del diritto del figlio a conoscere la sua origine»: con la conseguenza, «in definitiva», che «lo squilibrio iniziale resta perpetuato nella misura in cui il diritto all'accesso alle informazioni sulle origini personali resta subordinato alla decisione esclusiva della madre» (così, Corte cassaz., *op. et loc. ult. cit.*, tredicesimo e quattordicesimo per.).

[64] Così, Corte cassaz., *op. et loc. ult. cit.*, quinto per.

[65] … mettendosi, insomma, «in luce come all'istituto del parto anonimo è stata riconosciuta nella sentenza *Odièvre* legittimità anche nel perpetuare una posizione di disparità fra gli interessi in conflitto rendendo per certi versi improprio il richiamo alla teoria e alla tecnica del bilanciamento fra diritti fondamentali abitualmente utilizzata dalla giurisprudenza di Strasburgo»: così, Corte cassaz., sez. I civ., sent. 21 luglio 2016, n. 15024 (punto 14 *cons. dir.*).

[66] … laddove, in altri termini, si «riconosce come un ostacolo assoluto a qualsiasi ricerca di informazione, da parte della persona nata in regime di anonimato, la decisione della madre, quale che sia la ragione e la legittimità di tale decisione»: così, Corte cassaz., *op. et loc. ult. cit.*, terzo per.

[67] Così, Corte cassaz., *op. et loc. ult. cit.*, settimo per.

Special issue

Coming without *coming from*: The adoptee's right of access to origins within the constraints of maternal anonymity

Stefano Agosta*

ABSTRACT: This article analyses the judicial path, crossed by lights and shadows, and its ability to make it feasible to more easily learn information about one's own parental history in anonymous birth. A comparison is made between the European Court of Human Rights (ECtHR) case law and the Italian Constitutional Court case law, showing differences in the methods but strong similarities in the substantive solutions. Conclusively, in the Italian legal system, the mother's decision to confirm her original choice for anonymity has an undisputed prevalence when it tries to balance with the child's constitutional right to have his or her own personal experience recognised.

KEYWORDS: Anonymous birth; right to know one's origins; right to respect for private life

SUMMARY: 1. Personal identity and knowledge of origins: introduction. – 2. A comparison between the mother's right not to be found and the adoptee's right to seek: a static perspective. – 3. (*continue*) The constitutional mosaic (between health, privacy and personal identity). – 4. (*continue*) The dynamic perspective. – 5. The centrality of the maternal veto and the residual ambiguities of a (disguised) balancing.

1. Personal identity and knowledge of origins: introduction

Among the many intersections that inevitably exist at present between law and genetics – for reasons that can be easily understood, depending on medical-scientific progress – during the last few years, a place in adjudication and doctrine has surely been carved out (and continued to hold strongly) by the right of the individual to research his genetic and biological origins.

There are at least two viewpoints from which the delicate issue of the knowledge of one's personal history can be looked at as a whole in our legal system – that is, respectively, from the perspective of powers or from the perspective of rights. Here, particular attention will be paid only to the second perspective,[1] with specific reference to the right to personal identity pursuant to Article 2 of the Italian Constitution, with the need to access one's past experience representing one of its most salient

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[1] … the first of these two viewpoints could be examined in greater detail on another occasion.

aspects.[2] It is no mystery that "the balanced development of the individual and relational personality" passes through the "construction of one's external identity, whose essential elements are the name and a recognisable descent" and, on the other hand, through the construction of a specular "internal" identity, requiring "the knowledge and acceptance of the biological descent and of the closest parental network".[3]

From this point of view, it is therefore obvious that the individual's relational life is profoundly affected by satisfying the innate need to learn information about his or her previous parental history.[4] This is true for the adopted child (to whom this contribution is expressly dedicated), but is all the more true for other situations that are to some extent comparable to the former. For example, that of the child born through heterologous insemination[5] or, backwards (and *in limine*), of supernumerary embryos that can potentially be adopted at birth.[6]

[2] In this sense, Italian Constitutional Court (ItCC in the following) no. 286/2016 (p. 3.4.1. *cons. dir.*, first subparagraph). See, *ex multis*, E. MALFATTI, *Illegittimità dell'automatismo nell'attribuzione del cognome paterno: la "cornice" (giurisprudenziale europea) non fa il quadro,* in *forum costituzionale* (5 January 2017); S. SCAGLIARINI, *Dubbie certezze e sicure incertezze in tema di cognome dei figli*, in *rivista AIC* (19 May 2017, available at: https://www.rivistaaic.it/images/rivista/pdf/2_2017_Scagliarini.pdf); C. INGENITO, *L'epilogo dell'automatica attribuzione del cognome paterno al figlio (Nota a Corte costituzionale n. 286/2016* (https://www.osservatorioaic.it/images/rivista/pdf/INGENITO%20definitivo.pdf) and A. FUSCO, *"Chi fuor li maggior tui?": la nuova risposta del Giudice delle leggi alla questione sull'attribuzione automatica del cognome paterno. Riflessioni a margine di C. cost. sent. n. 286 del 2016* (https://bit.ly/3hmtg3g), both in www.*osservatorioaic*.it (respectively 31 May e 5 September 2017).

[3] In this direction, Italian Court of Cassation, sec. I civ., judgment 29 May 2017-20 March 2018, no. 6963 (respectively, pp. 8 and 8.1, first subparagraph, *cons. dir.*), with notes by, *ex multis*, G. VASSALLO, *Parto anonimo: diritto di conoscere le proprie origini va esteso alle sorelle*, in www.altalex.com (12 April 2018); E. CATALANO, *Il diritto alla conoscenza delle proprie origini*, in www.salvisjuribus.it (4 July 2018); A. GIURLANDA, *Il diritto a conoscere le proprie origini può essere esercitato anche nei confronti delle sorelle e dei fratelli biologici dell'adottato?*, in www.questionegiustizia.it (26 September 2018); G. CASABURI, *Riflessioni estemporanee su azioni di stato, nuova genitorialità, tutela del minore,* en attendant *le SS.UU. del 6 novembre 2018*, in www.articolo29.it (8 November 2018); C. GRANATA, *Il diritto alla ricerca delle proprie origini: i punti rimasti irrisolti dopo la sentenza n. 6963 della Corte di Cassazione, Sez. I, del 20.03.2018*, in www.rivista.camminodiritto.it (16 December 2019); I. LOMBARDINI, *Il procedimento di "interpello" della madre biologica, che abbia dichiarato di non voler essere nominata al momento del parto, ai fini dell'eventuale revoca dell'originaria dichiarazione, e la progressiva espansione del diritto dell'adottato alla conoscenza delle proprie origini biologiche ad opera della recente giurisprudenza*, in www.diritto.it (5 June 2020).

[4] In this sense, ItCC no. 278/2013 (p. 4 *cons. dir.*, eight subparagraph) with notes of, *ex plurimis*, E. FRONTONI, *Il diritto del figlio a conoscere le proprie origini tra Corte EDU e Corte costituzionale. Nota a prima lettura sul mancato ricorso all'art. 117, primo comma, Cost., nella sentenza della Corte costituzionale n. 278 del 2013* (https://www.osservatorioaic.it/images/rivista/pdf/contributo%20Frontoni.pdf) and A. RAPPOSELLI, *Illegittimità costituzionale dichiarata ma non rimossa: un "nuovo" tipo di sentenze additive?* (https://bit.ly/2Qajbvh), both in www.osservatorioaic.it (respectively, December 2013 and January 2015).

[5] "In this personal dimension", in fact, "the possibilities offered by PMA techniques, known as "heterologous", solve medical problems, but profoundly modify parenthood and complicate questions about the search for one's origins'": in this sense, for example, V. DE SANTIS, *Diritto a conoscere le proprie origini come aspetto della relazione materna. Adozione, PMA eterologa e cognome materno*, in www.nomos-leattualitaneldiritto.it (March 2018), spec. 1. On the other side, it is precisely "the entry of 'donors' on the scene" that raises "the serious question of the human right to know one's origins, a question that cannot be overlooked, minimised or crushed by the weight of technical and health aspects": M. CASINI, C. CASINI, *Il dibattito sulla PMA eterologa*

As a peculiar aspect of the broader and multifaceted right to personal identity – to return to the specific situation of adoption – in recent years, the right to search for one's origins has benefited from a more or less broad recognition both at the international level and, as far as we are concerned, at the domestic level. The Italian discipline, which has been the result of a series of gradual (but not always orderly) stratifications over time,[7] has nonetheless reached the harsh result of denying the adoptee any authorisation to know his or her personal history – and to anyone interested in it, this cannot occur until a hundred years have elapsed since the certificate of assistance in childbirth or the medical records containing the identification data of the mother – without even contemplating a prior verification of the mother's persistent desire to remain anonymous.

The legal regulation was so stringent that it was foreseeable (even, inevitable) that the courts would intervene, in search of a more reasonable balance between the interests involved – "that of the person who wants to complete the construction of his identity through the search of his biological origins and that of the biological mother who has exercised, at the time of birth, the right not to be named and who may want to keep this secret precisely in order not to alter the identity, also relational, built over time".[8] Before looking dynamically at the constitutional issues involved, we first conducted an in-depth static analysis of the "right to know the truth about one's personal history" and "the right to preserve the pre-existing construction of one's own identity and that of any third parties involved".

Let us leave aside for a moment the purely synchronic profile of the two rights evoked (related, that is, to the need to guarantee the conceived and pregnant woman the best conditions for the birth

all'indomani della sentenza costituzionale n. 162 del 2014. In particolare: il diritto a conoscere le proprie origini e l'"adozione per la nascita", in this *review*, no. 2/2014, 139.

[6] Obviously, it is not possible here to dwell at length on this main issue. It is only possible to point out that "during the discussion on Law 40/2004, 'adoption for birth' or 'prenatal adoption' was proposed as a limited and temporary remedy, on the assumption that when the new law came into force, the accumulation of spare embryos in freezers would stop": but it was harshly rejected, at the end, by those who "saw in the 'declaration of adoptability of the conceived' the equating of the unborn with the already born" (thus, once again, M. CASINI, C. CASINI, op. cit., 151). In the years that followed, this proposal made a comeback thanks to a series of interventions by the National Bioethics Committee, which on several occasions highlighted the validity of the arguments [see part. Adoption for birth of cryopreserved and residual embryos resulting from medically assisted procreation (P.M.A.) and fate of embryos resulting from medically assisted procreation that can no longer be implanted, both at http://bioetica.governo.it, 18 November 2005 and 26 October 2007 respectively].
On this subject, see for instance A. PALAZZO, *La filiazione*, Milan, 2007, part. 52 ss.; M. PICOZZI, F. NICOLI, V. VIGANÒ, *Il dono tra desiderio e ragione. Una riflessione sui principali nodi bioetici connessi alla fecondazione eterologa*, in AA.VV., *Cose o persone? Sull'esser figli al tempo dell'eterologa*, edited by L. Grion, Trieste, 2016, spec. 58 ss.; D. CASTELLANO, *Congelamento degli embrioni: un caso e molti problemi*, in www.filodiritto.com (15 December 2020).

[7] In this sense, see the original wording of Art. 28 of law no. 184/1983, *Right of the child to a family*, as well as the subsequent amendments introduced, respectively, by Art. 30(1) (*Declaration of birth*) of Presidential Decree no. 396/2000, *Regulations for the revision and simplification of the civil status system, pursuant to Article 2(12) of law no. 127 of 15 May 1997*, by Art. 24 of law no. 149/2001, *Amendments to law no. 184 of 4 May 1983, on the adoption and foster care of children, and to Title VIII of the first book of the Civil Code*, and by Art. 93 (2) (*Certificate of birth assistance*) of Legislative Decree no. 196/2003, *Personal Data Protection Code*.

[8] Here again Court of Cassation, sec. I civ., judgment 29 May 2017-20 March 2018, no. 6963 (p. 8.1, *cons. dir.*, respectively, second and first subparagraph) to which we refer also for the textual passage immediately following.

and, in this way, avoid her assumption of irreversible choices[9]), and let us now focus on the diachronic profile (relating to the time after the birth). It is natural, in fact, that "the commitment to the recognition of the right to know one's origins has been stimulated, in very recent times, precisely by the need to find a balanced composition between opposing rights".[10]

2. A comparison between the mother's right not to be found and the adoptee's right to seek: a static perspective

It is easy to understand how there is, *in fact,* a strong reciprocal conditioning between the fundamental constitutional requirements underlying the rights of mothers and children respectively, because they are not even artificially separable *in abstract.*[11]

If we think of the mother, in particular, the law is aimed at preventing her from retracing her steps many years later – answering to an unknown and perhaps already grown-up child – by establishing that her original choice of anonymous childbirth was irreversible.[12] The legislator of the time had, in short, staked everything on the inextricable interweaving between the maternal right to anonymity and the non-breakability of secrecy.[13] Indeed, this solution did not seem to be a real balancing of opposing constitutional interests (which, chronologically, would have occurred, *in limine*, only when the

[9] See ItCC no. 278 cit. (p. 4, *cons. dir.*, fourth subparagraph, first indent), which, in particular, refers to the same passage in its own precedent no. 425/2005 (p. 4, *cons. dir.*, third subparagraph) [with notes of, *ex multis*, S. MARZUCCHI, *Dei rapporti tra l'identità dell'adottato e la riservatezza del genitore naturale (in margine alla sent. n. 425 del 2005 della Corte costituzionale)*, in www.associazionedeicostituzionalisti.it (6 April 2006); S. FAVALLI, *Parto anonimo e diritto a conoscere le proprie origini: un dialogo decennale fra CEDU e Corte Costituzionale italiana*, in www.forumcostituzionale.it (9 December 2013); B. BARBISAN, *Apprendimento e resistenze nel dialogo fra Corte costituzionale e Corte di Strasburgo: il caso del diritto all'anonimato della madre naturale*, in www.dirittticomparati.it (9 May 2016)].

[10] Here again Court of Cassation, sec. I civ., judgment 29 May 2017-20 March 2018, no. 6963 (p. 8.1, *cons. dir.*, second subparagraph).

[11] As has been pointed out by ItCC no. 278, cited above (p. 4 *cons. dir*, first and second subparagraphs), "the issue of the mother's right to anonymity and the child's right to know his or her origins for the purpose of protecting his or her fundamental rights have already been the subject of rulings both by this Court and by the European Court of Human Rights": since they are "issues of particular delicacy, because they both involve constitutional values of primary importance and see their respective ways of realising them mutually implicated"; "to the point that - as is evident - the scope of the protection of the mother's right to anonymity cannot but condition, in practice, the fulfilment of the child's opposing aspiration to know his or her origins, and vice versa".

[12] "The irrevocability of the effects of this choice was", in other words, "explained according to a logic of reinforcing the corresponding objectives, excluding that the decision for anonymity could entail, for the mother, "the risk of being, in an unspecified future and at the request of the child never known and already an adult, called upon by the judicial authority to decide whether to confirm or revoke that distant declaration of will"": thus, ItCC no. 278 cit. (*ibid.*, fourth subparagraph, second indent).

[13] ... "the founding nucleus of that choice" being "in this way, as easily understandable, in the bi-univocal correspondence between the right to anonymity, considered in itself, and the lasting and binding protection of confidentiality or, if you like, of secrecy, which the exercise of that right inevitably involves": thus, again, ItCC no. 278, cit. (*ibid.*, fifth subparagraph) where, in particular, it goes on to consider this last protection "a founding nucleus which – it is worth pointing out – cannot but be reaffirmed, precisely in the light of the values of primary importance which it intends to preserve".

woman had decided to opt for anonymous childbirth), since the possibility of deciding whether to maintain or revoke that original choice would have been open only for the mother.[14]

The mother's constitutional right to anonymity, therefore, seemed to prevail over the other rights at stake, but the reasons for it prevailing did not seem conclusive in the end. The need to exercise a right to be forgotten without external interference did not seem decisive, nor did the need to prevent legal proceedings aimed at ascertaining whether the wish to remain anonymous was still in force, such a proceeding jeopardising the secrecy of her identity.[15] These two reasons were not decisive because the adoptee's right to access his or her past would have been irreparably compromised, and this right is no less fundamental than the mother's right to be forgotten. Secondly, the effective guarantee of the woman's privacy would have depended on the introduction of an abstract possibility and a concrete way of questioning her.[16] The real point was, in reality, a possible reconsideration of the assumption of a parental status that is no longer legal but natural: the fact that the mother initially denied her legal parental status cannot exclude the fact that she may later accept and desire her natural parental status. In the end, the mother could reconcile her original renunciation of her legal parental status with a later acceptance of her natural parental status.[17]

[14] "Only the mother therefore in this perspective can be the person entitled to decide whether to revoke her decision to remain anonymous in relation to the breaking of that need for protection that allowed her to make the choice allowed by the law": thus, Court of Cassation, sec. I civ, judgment of 21 July 2016, no. 15024 (p. 15 cons. dir.) [with notes of, *ex multis*, G. NALIS, Osservatorio di diritto civile, in www.ildirittoamministrativo.it (28 February 2017); A. GIURLANDA, *op. cit.*; I. LOMBARDINI, *Una questione problematica ancora aperta dopo le recenti pronunce della giurisprudenza: il diritto dell'adottato, non riconosciuto alla nascita, alla conoscenza delle proprie origini e il diritto della madre biologica all'anonimato*, in www.diritto.it (6 April 2020) and ID., *Il procedimento di "interpello" della madre biologica*, cit.], sharing the opinion that "according to which, in this case, the balancing of the fundamental rights at stake appears to be an ineffective and in some ways inappropriate category (...)": being, in other words, "properly to speak of a balancing between fundamental rights" only "with reference to the moment of the mother's choice to give birth anonymously" – "because at this moment her right to life and that of her child are at stake" – and not already "after the birth", when "it is no longer the right to life that is at stake and the right to anonymity becomes instrumental in protecting the choice made from the social consequences and in general from the negative consequences that would primarily affect the mother" (on this crucial point, however, we will return, *infra*, to par. 5, at the end of this article).

[15] The reference would be, thus, to that "system" which – "making the space of the "constraint" to anonymity temporally equivalent to a duration that could exceed that of an ideal human life" – "rests on the need to prevent any injury to the "right to be forgotten" of the mother and, at the same time, the need to safeguard *erga omnes* the confidentiality of her identity, which was obviously considered at risk every time contact is sought to ascertain whether or not she intends to maintain her anonymity": thus, ItCC no. 278 cit. (p. 5, fourth subparagraph).

[16] In other words, none of the above-mentioned requirements could be said to be truly "diriment": not the first one, since the danger of disturbance to the mother corresponds to an opposing danger for the child, deprived of the right to know its origins; not the second one, since the greater or lesser extent of the protection of confidentiality remains, in conclusion, entrusted to the different modalities provided for by the relevant rules, as well as to the practise of their application": thus, again, ItCC no. 278 cit. (*ibid.*, fifth subparagraph).

[17] ... "on a more general level, a choice for anonymity entailing an irreversible renunciation of 'legal parenthood'" could "reasonably not imply", in other words, "also a definitive and irreversible renunciation of 'natural parenthood'": thus, once again, ItCC no. 278 cit. (*ibid.*, sixth subparagraph) where, in particular, it is admitted that "if this were the case, on the other hand, a sort of prohibition would be introduced into the system which would preclude any possibility of a reciprocal relationship between mother and child, with outcomes that would be difficult to reconcile with Article 2 of the Constitution". (*ibid.*, seventh subparagraph).

Turning to the child's circumstances, it is useful and necessary to distinguish the profile of the effective enforceability of the right to learn about his or her previous parental history from the profile related to the constitutional parameters.

It should be noted that constitutional caselaw at first ruled out infringement of Article 3 of the Italian Constitution on the principle of equal treatment, due to differing regulations regarding the right for an adopted child to seek their own origins, whether they are the adopted child of a mother seeking anonymity or one whose parents never expressed any opinion on the matter (i.e., the search for one's origins was excluded in the first case and permitted in the second);[18] "only the first" of the above circumstances would be "characterised by the conflictual relationship between the adopted child's right to his or her own personal identity and the mother's right to respect for her wish to be anonymous", "and not also the second".[19] Thus, the difference in legal treatment of the two cases appears reasonable.[20]

With reference, then, to the possible limitation of the right in question – especially when the applicant is already an adult – it can be easily argued that "the vital interest of the individual to obtain the information necessary to discover the truth with regard to an important aspect of [his] personal identity, as an integral part of the right to private life", integrates "a subjective and ultra-personal right and, therefore, its enforceability has no temporal limitation".[21] Moreover, well-established case law confirmed recently[22] – has recognised that the full guarantee of the right to personal identity also depends on the recognition of the right "to a filial 'status' corresponding to the biological truth".[23]

[18] ... that is to say, "from the point of view of the unreasonable difference in treatment between the adopted child born of a woman who has declared that she does not wish to be named and the adopted child of parents who have not made any declaration and have, on the contrary, undergone the adoption". This legislative decision could have been considered unreasonable, "prohibiting the former from accessing information on his or her origins while allowing the latter to do so. The balance between the adoptee and his or her adoptive parents [could have been] exposed to greater dangers in the latter case than in the former, where years later the biological parent could have worked out his or her past conduct", ItCC no. 425, cit. (p. 6, *cons. dir.*, first subparagraph).

[19] In this sense, ItCC no. 425 cit. (*ibidem*, second subparagraph).

[20] The ItCC overruling of this decisive aspect will be discussed *infra* in section 4.

[21] In this way, European Court of Human Rights (ECtHR in the following), sec. II, *Godelli v. Italy*, 25 September 2012, § 54, with comments of, *ex plurimis*, D. BUTTURINI, *La pretesa a conoscere le proprie origini come espressione del diritto al rispetto della vita privata*, in www.forumcostituzionale.it (24 October 2012); R.G. CONTI, *La giurisprudenza civile sull'esecuzione delle decisioni della Corte Edu*, in www.questionegiustizia.it, no. 1/2019, 283 ss.; R. TREZZA, *Diritto all'anonimato e diritto a conoscere le proprie origini biologiche*, in www.giustiziainsieme.it (4 October 2019); I. LOMBARDINI, *Il procedimento di "interpello" della madre biologica*, cit.

[22] Here Court of Cassation, sec. I civ., judgment 22 September 2020, no. 19824 with notes of, for example, S. OCCHIPINTI, *Accertamento della maternità, il diritto della madre all'anonimato cessa con la sua morte*, in www.altalex.com (2 October 2020); REDAZIONE, *Diritto a conoscere le proprie origini*, in www.diritto.it (8 October 2020); L. BONARINI, *Azione giudiziale di accertamento della maternità – parto cd. anonimo. Cass. Civ., sez. I, 22/09/2020, n. 19824*, in www.salvisjuribus.it (27 November 2020), and now please allow for reference to S. AGOSTA, *Anonimato della madre premorta e riespansione del diritto all'identità personale del figlio (a margine di Cassaz. sent. n. 19824/2020)*, in *Quad. cost.*, 2021.

[23] ... "the uncertainty on such a 'status'" could "determine distress and a 'vulnus' to the adequate development and formation of the personality in every stage of life": with the consequence that "the right to the recognition of a filial status corresponding to the truth belongs to the core of each person's inviolable rights (Art. 2 It. Cost.

The Italian legal system has given preeminent importance to this last right – "as an essential component of the right to personal identity, at every stage of a person's life and therefore also in adulthood".[24] This is, moreover, amply demonstrated by the lack of any temporal constraint on the legitimate activation by the interested person of the judge's verification of parenthood.[25]

3. (*continue*) The constitutional mosaic (between health, privacy and personal identity)

Let us now speak of constitutional parameters. There have been three main provisions that have traditionally underpinned the right in question: Article 32, Article 117(1), and Articles 2 and 3 of the Italian Constitution, depending on which one was invoked.

Let us start with the first provision, Article 32 of the Italian Constitution, concerning the right to psycho-physical health. This right has been put into play both in relation to the disclosure of the secret and in the diametrically opposed hypothesis of its maintenance. On the one hand, it has been argued, for example, that the judge (in this case, the European Court of Human Rights, also the Strasbourg Court or ECtHR in the following) should have taken due account of the harm to the psycho-physical well-being of the person adopted at a tender age (and, at the time of the appeal, already elderly) that might have resulted from the judicial removal of the anonymity.[26] On the other hand, however, it was objected that the appellant herself had "demonstrated a genuine interest in knowing the identity of the mother, since she had attempted to acquire certainty in this regard": "such behaviour" demonstrated "a moral and psychological suffering, even if this is not ascertained from a medical point of view".[27] The same parameter is relevant considering the potential violation of Article 32

and Art. 8 ECHR), considered both in the individual and relational dimension" [thus, Court of Cassation, sec. I civ., judgment 22 September 2020, no. 19824 (p. 2, *cons. dir.*, sixth subparagraph) recalling among others, on this point, Court of Cassation, sec. I civ., judgment 13 April-9 June 2015, no. 11887; 29 November 2016, no. 24292; 15 February 2017, no. 4020]. "On the grounds of these articulated arguments", the Court of Cassation, moreover, held "the question about the constitutional legitimacy of Art. 270 of the Civil Code" to be manifestly unfounded. The question complained "that the action for the judicial ascertainment of paternity or maternity could not be time barred, [excluding] any possibility for the judge to assess the request for judicial declaration in cases where the action [had been] proposed with considerable delay (in this case about forty years), with the effect of sacrificing the right of the presumed father to the stability of family relationships matured over time, and imposing on him after a long time a compulsory ascertainment of the filiation relationship that the interested person could have requested earlier": so, again, Court of Cassation, sec. I civ., judgment 22 September 2020, no. 19824 (*ibid*.).

[24] Again, Court of Cassation, sec. I civ., judgment 22 September 2020, no. 19824 (*ibid.*, seventh subparagraph).

[25] ... "as well as the fact that evidence may be given by any means, pursuant to Art. 269, par. 2 of the Civil Code": Court of Cassation, *op. et loc. ult. cit*.

[26] "According to the Italian Government", in particular, "the Court" should precisely have "taken into account the fact that the applicant, now almost 70 years old, was adopted at the age of six and that the non-consensual lifting of the secrecy of her birth [could] have proved very difficult at this stage, given the possible non-negligible risks to her health and to her present family": see ECtHR, *Godelli v. Italia*, cit., § 58.

[27] If it was, therefore, realistic to think "that the applicant, [at the time] 69 years old, [had] managed to build up her personality even in the absence of information concerning the identity of her biological mother, it [had to be] accepted that the interest that [an] individual might have in knowing her ancestry did not [diminish] with age, indeed [the] opposite occurred": thus, again, ECtHR, *Godelli v. Italy*, cited above, § 69, on the point recalling - among its precedents on the matter – EctHR, third section, *Jäggi v. Switzerland*, 13 July 2006, spec. § 40, with comments, *ex multis*, of C. CAMPIGLIO, *Con la morte, l'uomo perde il diritto al rispetto della vita privata*

when the child found herself in the material impossibility of accessing any information relating to her parents' genetic makeup.[28]

As for the second relevant constitutional provision, Article 117(1) has often been invoked in connection with Article 8 of the European Convention on Human Rights (ECHR) and the corresponding interpretation offered by the Strasbourg judges, together with the New York and Hague Conventions.[29] In the case of *Godelli v. Italy* cited above, the Strasbourg Court sanctioned the Italian discipline for not having "sought to establish a fair balance and proportionality between the interests of the parties to the dispute" and, in so doing, for having consequently "exceeded the margin of discretion which [it] had been granted [by the ECHR]".[30] On the one hand, it was held that the need to know one's personal history could fall within the concept of both private and family life (both of which are protected by Article 8 ECHR).[31] On the other hand, however, the ECtHR clearly limited the case to private life only. Some of the Court's famous precedents on the matter have been quoted[32] to demonstrate that, in fact, the application was made only for access to the identity of one's biological ances-

and S. TONOLO, *Identità personale, maternità surrogata e superiore interesse del minore nella più recente giurisprudenza della Corte europea dei diritti dell'uomo*, both in *Diritti umani e diritto internazionale*, respectively no. 2/2007, 394 ss. and no. 1/2015, 202 ss.; L. POLI, *Il diritto a conoscere le proprie origini e le tecniche di fecondazione assistita: profili di diritto internazionale*, in *GenIUS*, no. 1/2016, 43 ss.

[28] ... "because of the impossibility", in other words, "for the child to obtain data on his family history, also in relation to the genetic risk": "inasmuch as preventing knowledge of data concerning the birth mother would deprive the adoptee of any possibility of obtaining an anamnesis of the family, which is essential for prophylactic interventions or diagnostic tests, since he of she already lacks information on the health history of the paternal branch of the family tree. This, moreover, in the light of the practice, widespread in Italian hospitals, of omitting the ordinary collection of anamnestic data that do not identify the mother" [ItCC no. 278 cited above (p. 1, *cons. dir.*, second subparagraph, and p.1 *rit. fatto*, eighth subparagraph)].

[29] … for "violation" of, respectively, "Artt. 7 and 8 of the New York Convention on the Rights of the Child of 20 October 1989, made enforceable by law no. 176 of 1991, in so far as they require respect for the rights of the child, including those aimed at preserving his or her identity, name and family relationships" ("for the adopted child, identity" consisting "precisely in seeking his origins, his roots and information about his biological family") as well as "of Art. 30 of the Hague Convention of 29 May 1993, made enforceable by law no. 476 of 1998": thus, Court of Cassation, sec. I civ., judgment 29 May 2017-20 March 2018, no. 6963 (p. 4.1, *cons. dir.*)

[30] Here ECtHR, *Godelli v. Italia*, cit., § 71 [§ 58 English version, n.d.r.], quoted by ItCC no. 278, cit. (p. 1. cons. dir., second subparagraph).

[31] … "the applicant" having in particular argued "that her request to obtain information on eminently personal aspects of her history and childhood [fell] within the scope of Article 8 of the Convention" since "the search for her identity [was] an integral part of her 'private life' but also of her 'family life'": thus, ECtHR, *Godelli v. Italia*, cit., § 43.

[32] Spec. *Mikulić v. Croatia*, 7 February 2002, § 53 [with comments of, *ex multis*, C. CAMPIGLIO, *Il divieto di fecondazione eterologa all'esame della Corte europea dei diritti umani*, in *Diritti umani e diritto internazionale*, no. 3/2010, spec. 4, D. BUTTURINI, *op. cit.*, part. 3, A. CIERVO, *Il diritto all'anonimato della madre biologica ovvero quando Strasburgo anticipa Roma*, in https://diritti-cedu.unipg.it/ (15 February 2014), par. 2] and *Odièvre c. Francia*, 13 February 2003, § 29 with notes of, *ex plurimis*, J. LONG, *Ammissibilità del parto anonimo e accesso alle informazioni sulle proprie origini: il caso Odièvre c. Francia (introduzione a Corte europea per i diritti dell'uomo, sentenza 13 febbraio 2003, Odièvre c. Francia)*, in *Minori e giustizia*, no. 3/2003, 172 ss.; A. RENDA, *La sentenza O c. Francia della Corte Europea dei diritti dell'uomo: un passo indietro rispetto all'interesse a conoscere le proprie origini biologiche*, in *Familia*, no. 4/2004, 1121 ss.; S. FAVALLI, *op. cit.*

tors (as an important component for the construction of one's personal identity) and not also for verification of one's adoptive status (which, on the contrary, falls within the notion of family life).[33]
Questioned on this point, the Strasbourg judges recalled "that Article 8 protects a right to personal identity and development and the right to establish and deepen relations with one's peers and the outside world": "to that development", contributes "the discovery of details relating to one's identity as a human being and the vital interest, protected by the Convention, in obtaining information necessary to discover the truth concerning an important aspect of personal identity, for example, the identity of one's parents".[34] From this last point of view, the Strasbourg Court has pointed out that, among the duties that Article 8 imposes on each national system, there is no clear demarcation between negative and positive obligations; on the contrary, the positive obligations – for instance, the need to adopt any measure aimed at ensuring the effectiveness of the protection of privacy – can overlap with the mere obligation of the State to abstain from any abusive public interference in the private sphere of the person.[35]

[33] "In the present case", in short, "the applicant was not seeking to question the existence of her adoptive filiation, but to know the circumstances of her birth and abandonment, which included knowledge of the identity of her biological parents": with the result that "the Court" was not "called upon to determine whether the proceedings concerning the filial link between the applicant and her mother fell within the scope of 'family life' within the meaning of Article 8, since in any event the right to know one's ancestry fell within the scope of the concept of 'private life', which included important aspects of personal identity of which the identity of the parents formed part" (thus, ECtHR, *Godelli v. Italia*, *ibid.*, § 45).
On this particular point, however, it is worth recalling that "the Government [had argued] that no family life within the meaning of Article 8 of the Convention [existed] between the applicant and her biological mother, as the former had never seen her mother, since the latter had never wanted to meet her and consider her as her child" ("in fact, she" having "expressly expressed her wish to abandon her" and "accepted that her daughter should be adopted"): "by guaranteeing the right to respect for family life, Article 8 presupposes", on the other hand, "the existence of a family (*Marckx v. Belgium*, judgment of 13 June 1979, Series A no. 31)"; "if the case-law did not [require] that there [be] cohabitation between the various members of the 'family', [there should] at least have been close personal relationships between them" which would have demonstrated "an affective relationship between two beings and their willingness to entertain that relationship would [have] been fundamental for the organs of the Convention" ("the latter" also considering "that the biological link alone [was] insufficient, in the absence of close personal ties between the persons concerned, to constitute a family life within the meaning of Article 8") (so, *ibid.*, § 44).
[34] With the consequence that "the birth, and in particular the its circumstances, is part of the private life of the child, and then of the adult, sanctioned by Article 8 of the Convention, which thus finds application in the present case": thus – also invoking the precedent set out in *Mikulic v. Croatia*, cited above, §§ 54 and 64 – ECtHR, *Godelli v. Italia*, cit., § 46, also referred to by Court of Cassation, sec. I civ., judgment of 21 July 2016, no. 15024 (p. 9, *cons. dir.*) where, in particular, it recalls how "the European Court of Human Rights (...) has given an interpretation of Article 8 ECHR, which places the right to knowledge of one's origins within the scope of the concept of private life and specifically within the sphere of protection of personal identity", "in this perspective", stating "that Article 8 protects the right to personal identity and fulfilment and the right to establish and develop relations with one's peers and the outside world".
[35] "[A]lthough the object of Article 8 is essentially that of protecting the individual against arbitrary interference by the public authorities, it does not merely compel the State to abstain from such interference: in addition to this primarily negative undertaking, there may be positive obligations inherent in an effective respect for private life. These obligations may involve the adoption of measures designed to secure respect for private life even in the sphere of the relations of individuals between themselves (see *X and Y v. the Netherlands*, 26 March 1985, § 23, Series A no. 91) ". Moreover, "[T]he boundaries between the State's positive and negative obligations under Article 8 do not lend themselves to precise definition. The applicable principles are nonethe-

The Italian Constitutional Court, intervening on the matter, formally declared the violation of Article 117(1) be absorbed, without ruling on the merits.[36] However, there are no doubts that the Court has incorporated what was already observed at the conventional level. This was evident when it was noted that there were different outcomes between the French and the Italian legislation with respect to the common need to open a – albeit minimal – window to the right for the adoptee to access his or her past.[37]

Let us now deal with Articles 2 and 3 of the Italian Constitution. First of all, considering method,[38] it must be remembered that the interpretation of the Italian regulation on anonymity must comply both with the Italian Constitution (in particular Articles 2, 24, and 30) and with the ECHR [pursuant to Article 117(1) quoted above].[39] Knowledge of the circumstances surrounding one's birth and subsequent separation from the mother, as well as the emotional relationships established, contribute to the full realisation of one's parental story. It is for this reason that the right to search for one's origins can certainly take root within the borders of constitutional provisions as well as ECHR provisions protecting private and family life.[40]

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less similar. In particular, in both instances regard must be had to the fair balance which has to be struck between the competing interests, and in both contexts the State enjoys a certain margin of appreciation (see *Mikulić*, cited above, § 58)" (ECtHR, *Godelli v. Italy*, cit., § 60 [§ 47 English version, n.d.r.], quoted by Court of Cassation, sec. I civ., judgment 22 September 2020, no. 19824 (p. 2, *cons. dir.*, sixteenth subparagraph) recalling that "Article 8 ECHR, as interpreted by the ECtHR (ECtHR, 22/09/2012, *Godelli v. Italy*, ECtHR, 13/02/2003, *Odievre v. France*), aims essentially at protecting the individual against arbitrary interference by the public authorities, not only ordering the State to refrain from such interference, but adding positive obligations inherent in effective respect for private life; these cannot but include the right to bring all the proceedings that the domestic legal system itself offers for the recognition of a person's status as a natural child".

[36] ItCC no. 278, cit. (p. 6, *cons. dir.*, seventh subparagraph).

[37] "This, in fact, is based on the same remarks, in substance, formulated by the ECtHR in the 'Godelli judgment'" when it "criticised the fact that the Italian legislation does not give "any possibility for an adopted child who is not recognised at birth to request access to non-identifying information on his origins or the reversibility of the secret", unlike what is provided for in the French system, which was examined in part in the judgment of 13 February 2003 in the 'Odièvre case'", judgment no. 278 cit. (*ibid*., second and third subparagraphs respectively).

[38] See, for example, *A. RAUTI, La "cerchia dei custodi" delle "Carte" nelle sentenze costituzionali nn. 348-349 del 2007: considerazioni problematiche, in AA.VV., Riflessioni sulle sentenze 348-349/2007 della Corte costituzionale, edited by C. Salazar and A. Spadaro, Milan, 2009,* spec. 310; G. ROLLA, *Il processo di ibridazione dei sistemi accentrati di giustizia costituzionale. Note di diritto comparato*, in AA.VV., *Estado constitucional, derechos humanos, justicia y vida universitariaEstudios en homenaje a Jorge Carpizo*, edited by M. Carbonell Sánchez, H. Fix Zamudio, L. Raúl Gonzálea Pérez, D.Valadés Ríos, Mexico, 2015, part. 529; P. COSTANZO, L. MEZZETTI, A. RUGGERI, *Lineamenti di diritto costituzionale dell'Unione europea*, Turin, 2019, spec. 287 ss.

[39] Here Court of Cassation, sec. I civ., judgment 22 September 2020, no. 19824 (p. 2, *cons. dir.*, sixteenth subparagraph).

[40] If "the constitutional and conventional framework of the right to know one's own origins, as a declination of primary importance of the right to personal identity, is given by Articles 2 and 3 of the Italian Constitution and Article 8 ECHR", there is no doubt that "the development of the individual personality and the harmonious conduct of one's private and family life require the construction of one's own individual identity, based not only on a recognisable affective-educational parental context, but also on information relating to one's own birth useful to reveal the secret and the reasons for the abandonment": Court of Cassation, sec. I civ., judgment 9 November 2016, no. 22838 (p. 4.1, *cons. dir*., first subparagraph).

4. (*continue*) The dynamic perspective

Moving on to the substantive level, it has been pointed out that the violation of Articles 2 and 3 of the Italian Constitution – with the subsequent constitutional obligation of removal – depends on the legislative imposition of absolute and unconditional maternal anonymity;[41] this being diametrically opposed to "the right to seek one's own origins and hence the right to personal identity of the adopted person", and also unreasonably discriminating "the adopted child born of a woman who has declared that she does not wish to be named and the adopted child of parents who have not made any declaration and have actually undergone the adoption".[42]

Within these premises, if we look again at the constitutional values at stake in a dynamic sense, there is no doubt that national legal systems have a wide margin of appreciation whenever they are called upon to "choose the means that they deem most suitable to ensure a fair balance between the protection of the mother and the legitimate request" to know one's personal history "having regard to the general interest"[43] (especially when there is not sufficient common ground between the European States on how to reconcile the opposing needs of two private individuals).[44] On the grounds of Article 8 ECHR, there are many factors conditioning the more or less extensive discretionary power of the State (including the specific circumstances of private life that are at stake in each concrete case).[45]

However, the domestic discretion does not preclude the judicial authority that is adopted by a subsequent control on legislative measures. Surely the ECtHR and the Constitutional Court differ in the methods of respective judgments. But in terms of substance, there are interesting argumentative overlaps between conventional and domestic rulings. Thus, for example, from a methodological point of view, the Strasbourg Court should not overlap with the local authorities in identifying the most appropriate instrument for regulating the anonymity of the mother, its review necessarily focusing on the specific case under examination without extending to the State regulation considered in the abstract and as a whole.[46]

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[41] In this sense ItCC no. 278, cit. (p. 6, *cons. dir.*, sixth subparagraph).

[42] Again ItCC no. 278, cit. (p. 1, *cons. dir.*, second subparagraph).

[43] ECtHR, *Godelli v. Italia*, cit., § 67 [§ 52 English version, n.d.r.]

[44] The Government submitted that "the State enjoyed a margin of appreciation in the event of a conflict between two private interests. That margin of appreciation was enlarged in the instant case by the fact that no European consensus on the issue of a child's access to information about its origins existed", in ECtHR, *Godelli v. Italia*, *ibid.*, § 59 [§ 46 English version, n.d.r.]

[45] The Strasbourg Court itself "reiterates that the choice of the means calculated to secure compliance with Article 8 in the sphere of the relations of individuals between themselves is in principle a matter that falls within the Contracting States' margin of appreciation. In this connection, there are different ways of ensuring respect for private life, and the nature of the State's obligation will depend on the particular aspect of private life that is at issue (see *Odièvre*, cited above, § 46). The extent of the State's margin of appreciation depends not only on the right or rights concerned but also, as regards each right, on the very nature of the interest concerned", ECtHR, *Godelli v. Italia*, *ibid.*, § 65, second and third indent [§52 English version, n.d.r.]

[46] "In cases arising from individual applications the Court's task is not to review the relevant legislation or practice in the abstract; it must as far as possible confine itself, without overlooking the general context, to examining the issues raised by the case before it ... Consequently, the Court's task is not to substitute itself for the competent national authorities in determining the most appropriate policy for regulating matters" (see *S.H. and Others v. Austria* [GC], no. 57813/00, § 92, ECHR 2011) of anonymous births. It is not for the Court to re-

However, an overlap of arguments emerges with regard to the substance of the decision, starting with the type of scrutiny required. The starting point is that the concept of private life cannot exist without the right to personal identity and this one, in turn, is connected to the right to access one's own experience, in a sort of Chinese box system. For this reason, the judicial review appears stricter in balancing the constitutional requirements at stake[47] and, ultimately, in determining whether the best balance has been struck between the mother's right to an anonymous birth and the child's right to information about his or her parental history, in the light of the given historical and contextual conditions;[48] "the right to identity, as an essential condition of the right to autonomy (*Pretty v. United Kingdom*) and development of the person (*Bensaid v. United Kingdom*)", in fact, "forms part of the hard core of the right to respect for private life and therefore a more rigorous examination is required in order to effectively balance the interests at stake".[49]

It is precisely this strict scrutiny that has led to quashing the maternal right to anonymous childbirth. Originally, the legislative solution seemed in compliance with Article 2 of the Italian Constitution as the expression of a correct balance between the requirements at stake.[50] But due to the unlimited and unconditional protection offered to the mother,[51] it ended up being unlawful, both by the ECtHR (2012) and the Italian Constitutional Court finding that followed shortly thereafter (2013). Both Courts, in fact, considered the rigidity of the law on anonymous childbirth to be entirely disproportionate, and therefore excessive.

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view the necessity of the absolute ban that was found constitutional by the Italian legislature, comparing the rights that are protected by the Convention, as long as this measure is not arbitrary and the balancing reasonably takes into consideration all the rights in question" [in this sense, dissenting opinion of Judge A. Sajó (*ibidem*), cit.].

[47] ECtHR, *Godelli v. Italia*, *ibid.* (forth subparagh) [§52 English version, n.d.r.].

[48] "In situations where the Convention rights of two right-holders come into conflict, the role of the Court is to satisfy itself that a proper balance has been struck in the case. This means that an appropriate margin of appreciation must be afforded to the domestic authorities to carry out the balancing exercise; the role of the Court is supervisory. Where the balancing exercise has been undertaken by the national authorities in conformity with the criteria laid down in the Court's case-law, the Court would require strong reasons to substitute its view for that of the domestic courts (see *Von Hannover v. Germany (no. 2)* [GC], nos. 40660/08 and 60641/08, § 107, ECHR 2012)" (in this sense ECtHR, *Godelli v. Italy*, cit., § 66 [in the English version dissenting opinion of Judge A. Sajó (*ibidem*), n.d.r.]. "The choice of the most appropriate means of ensuring, in a fair manner, the reconciliation of the need for protection of the mother, who is in such a difficult position as to prevent her from assuming her parental role, with the legitimate demand of the child to have access to information about his or her origins is a matter for", in other words, "the States parties to the Convention": "however, the Court is in a position to review the choice and the actual exercise of those means of settling the conflict and, in particular, the search for and achievement of a balance between the competing interests and rights at stake": thus, Court of Cassation, sec. I civ., judgment 21 July 2016, no. 15024 (p. 11, *cons. dir.*).

[49] Again Court of Cassation, sec. I civ., judgment 21 July 2016, no. 15024 (p. 13 *cons. dir.*, eighth subparagraph).

[50] ItCC no. 425, cit. (*ibidem*, fifth subparagraph).

[51] This is because, as seen before, "the choice of the pregnant woman in difficulty that the law wishes to favour – in order to protect both her and the unborn child – would be rendered extremely difficult if the decision to give birth in an appropriate medical facility, remaining anonymous, could entail for the woman, on the basis of the same rule, the risk of being, in an unspecified future and at the request of a child she has never known and who is already an adult, questioned by the judicial authority to decide whether to confirm or revoke that distant declaration of will": thus, ItCC, judgment no. 425 cit. (p. 4, fourth subparagraph).

According to the Strasbourg judges, the violation of Article 8 ECHR comes immediately from the unlimited prevalence of the mother's need to preserve her anonymity over the competing interest of the child in the search for her origins, without the Italian legislature having made any effort to identify a minimum regulatory solution, so to speak, to redress the balance provided, as was done by the corresponding French legislation.[52] "… [W]here the birth mother has decided to remain anonymous, Italian law does not allow a child who was not formally recognised at birth and was subsequently adopted to request either access to non-identifying information concerning his or her origins or the disclosure of the mother's identity": and consequently, "the Italian authorities failed to strike a balance and achieve proportionality between the interests at stake and thus overstepped the margin of appreciation which it must be afforded".[53]

5. The centrality of the maternal veto and the residual ambiguities of a (disguised) balancing

The Italian Constitutional Court, on the other hand, quashed the Italian legislation for the absoluteness of its provisions, focusing on the existence of a real "crystallisation" or "immobilisation"[54] in the application of the right to maternal anonymity.[55] Since it was exercised by the mother, the right to anonymous childbirth ended up, on the one hand, becoming an irreversible legal obstacle for the child who wished to know his or her personal history but, on the other hand, turning into a sort of real "expropriation" to the detriment of the woman herself; from that moment on, she would, against her will, find herself deprived of any alternative option by the very system which, in theory, intended to protect her.[56]

The jurisprudential path that we have briefly tried to describe in these pages has been troubled, but in the end the ordinary courts, and their living interpretation, identified gaps in the narrow regulatory mesh of the right to maternal anonymity. It was, for example, accepted that a judicial action to es-

[52] Indeed, "The Court notes that, unlike the French system examined in *Odièvre*, Italian law [did] not attempt to strike any balance between the competing rights and interests at stake. In the absence of any machinery enabling the applicant's right to find out her origins to be balanced against the mother's interests in remaining anonymous, blind preference [was] inevitably given to the latter. Moreover, in *Odièvre* the Court observed that the new [French] law of 22 January 2002 improved the prospect of obtaining agreement to waive confidentiality and would facilitate searches for information about a person's biological origins as a National Council for Access to Information about Personal Origins had been set up. The law was of immediate application and now allowed the persons concerned to request disclosure of their mother's identity, subject to the latter's consent being obtained (see *Odièvre*, cited above, § 49), and to have access to non-identifying information", in ECtHR, *Godelli v. Italy*, cit., § 70 [§57 English version, n.d.r.].

[53] ECtHR, *Godelli v. Italy*, cit., § 71 [§58 English version, n.d.r.].

[54] In this sense ItCC no. 278 cit. (p. 5, *cons. dir*., second subparagraph).

[55] In this sense ItCC no. 278 cit. (p. 6, *cons. dir*., first subparagraph).

[56] "Once the choice for anonymity has been made, in fact, the relevant manifestation of will assumes connotations of irreversibility intended, substantially, to 'expropriate' the person holding the right from any further option; ultimately, the right is transformed into a sort of compulsory constraint, which ends up by having an expansive effect outside its holder and, therefore, by projecting the impediment to its eventual removal onto the child itself, to whose position it was originally intended to link the obligation of secrecy on the person who generated it": thus, ItCC no. 278, cit. (p. 5, second subparagraph).

tablish the link with the woman could be legitimately brought when she had in fact disavowed her original decision to abdicate her legal maternity. However, outside of such cases, the protection of maternal secrecy regarding childbirth remains maximal and lasting.[57]

In light of the above, in short, the undisputed prevalence of the right to anonymous childbirth – for the entire span of the woman's existence – seems to have been fully confirmed in recent times, every time it comes into the constitutional balance with the competing right of the child to have his or her own personal experience recognised; this is evidently still based on the future risk of disclosure of the mother's decision during the delicate moment of birth and which could push her to make irreversible decisions for the health and life of herself and the unborn child she is carrying.[58]

In the light of these considerations, the Italian attempt to make more flexible the original preclusion to learn information of one's own parental history appears to be crossed by lights and shadows, and it is considered alongside the French experience (where this result was achieved through legislation). Thus, for example, the perplexities already emerged in the Strasbourg case law on the French case, both in terms of method and merit, could be extended without too much difficulty to the Italian experience.[59]

Allowing a reconsideration of the initial choice for anonymity – the Italian ordinary courts have undoubtedly determined, on the one hand, a timid opening in favour of the adoptee's right to the search of his or her origins (as happened in parallel in France with the often cited law no. 93/2002, *relative à l'accès aux origines des personnes adoptées et pupilles de l'Etat).*[60] However, already on the merits, in Italy there is no doubt that "such reversibility [is] ultimately entrusted to and conditioned by the agreement" of the mother, the latter "[being] only invited and not [having] the obligation to

[57] … In other words, "this rule" could be "at the limit, derogated (thus allowing the exercise of the judicial ascertainment of maternity) only if it was the mother (...) with her own unequivocal conduct, to have shown the will to revoke in fact the choice, taken at the time, of renouncing legal parenthood, welcoming in her home the child as a son": "however, outside the borderline case set out above, the protection of the mother's right to anonymity, for the duration of her life, must be, as said, maximal" [thus, Court of Cassation, sec. I civ., judgment 22 September 2020, no. 19824 (p. 2, *cons. dir.*, respectively, tenth and eleventh indents)].

[58] "In the balancing of constitutional values that the interpreter must respect, facing the right to recognition of the status of parenthood, the mother's right to remain anonymous at the time of childbirth is in any case in a pre-eminent position": "This latter right, in fact, (...) is aimed at protecting the supreme goods of health and life, as well as that of the unborn child, of the mother, who could be induced to make choices of a different nature, a source of possible great risk for both, if, at the moment of extreme fragility that characterises childbirth, the woman who opts for anonymity has only the doubt of being exposed, subsequently, to an action of judicial ascertainment of maternity": so, again recently, Court of Cassation, sec. I civ., judgment 22 September 2020, no. 19824 (*ibid.*, respectively, eighth and nineth subparagraph).

[59] "The above-mentioned judgment of the European Court *Odièvre v. France*, of which the *Godelli v. Italy* judgment is the consistent reaffirmation" already represented, moreover, "a painful precedent because it was pronounced at the end of a difficult search for a balance between very different legal traditions and positions of principle, as is eloquently represented in the dissenting opinion of judges Wildhaber, Bratza, Bonello, Loucaides, Cabral Barreto, Tulkens and Pellonpää": see Court of Cassation, sec. I civ., judgment 21 July 2016, no. 15024 (p. 13, *cons. dir.*, first subparagraph).

[60] The French legislation submitted to the ECHR's scrutiny "clearly recognises the need to strike a balance between the conflicting rights": "although it does not call into question the institution of *accouchement sous x*, it certainly marks a step forward in terms of access to knowledge of one's origins in that it makes it possible to call for the reversal of the secrecy regarding the mother's identity" (Court of Cassation, *op. et loc. ult. cit.*, nineth and tenth subparagraphs).

provide identifying information".[61] Legally, it is thus inevitable that this unilateral maternal decision can be opposed by the person concerned[62] (or removed by a third party).[63] Moreover, on a psychological level, there is little doubt that the woman ends up viewed as having "the purely discretionary right to bring a child into the world, placing it in a state of suffering and condemning it for life to ignorance of its origins".[64]

In addition to these considerations, one aspect of this method has attracted the most criticism from scholars. It has been critically observed – in the aforementioned *Odièvre* case (but the reflections are fully extendable, as we have seen, also to the current Italian experience) – that it is not possible to speak of a reasonable weighing up of all the constitutional requirements at stake, since the persistent validity of the discipline on maternal anonymity continues to maintain a basic inequality.[65] In conclusion, when it is accepted – irrespective of the reasons underlying it – that the mother's choice may constitute an insurmountable barrier for the adoptee on the road to knowledge of his or her personal history,[66] there is an inevitable risk that a blatant imbalance between all the needs involved will continue to be perpetrated (by law in France and by case law in Italy); "the pure and simple right of veto granted to the mother [imply] that the rights of the child recognised in the general system under the Convention (*Johansen v. Norvège*, *Kuzner v. Germany*), [are] entirely denied and forgotten".[67]

[61] ... as in the French legal system where, on the other hand, she "may always oppose her identity being revealed even after her death (Article L. 147-6 of the *'code de l'action sociale et des familles'* introduced by Article 1 of the law of 22 January 2002)": thus, Court of Cassation, *op. et loc. ult. cit.*, eleventh and twelfth indents.

[62] … "the mother's refusal" imposing itself, in fact, "on the child who has no legal means of opposing her unilateral will": thus, Court of Cassation. *op. et loc. ult. cit.*, forth indent.

[63] Such as, precisely, in France where it has not been "provided that the established National Council (nor any other independent body) can take a final decision on the removal of the secret, in view of the conflicting interests, when the mother confirms her refusal, definitively depriving the child's right to know his or her origin": with the consequence, "ultimately", that "the initial imbalance remains perpetuated to the extent that the right to access information on personal origins remains subject to the exclusive decision of the mother" (thus, Court of Cassation, *op. et loc. ult. cit.*, thirteenth and fourteenth indents).

[64] Here Court of Cassation, *op. et loc. ult. cit.*, fifth indent.

[65] ... highlighting, in short, "how to anonymous childbirth has been recognized in the judgment *Odièvre* legitimacy also in perpetuating a position of inequality between the conflicting interests, making in some ways improper the reference to the theory and technique of balancing fundamental rights habitually used by the case law of Strasbourg": so, Court of Cassation, sec. I civ., judgment 21 July 2016, no. 15024 (p. 14, *cons. dir.*).

[66] ... where, in other words, "the decision of the mother, whatever the reason and legitimacy of that decision, is recognised as an absolute obstacle to any search for information on the part of the person born anonymously": Court of Cassation, *op. et loc. ult. cit.*, third indent.

[67] In this way Court of Cassation, *op. et loc. ult. cit.*, seventh indent.

Special issue

Biotechnologies, Birth and the Right to Know One's Genetic Origins

Lucia Busatta, Simone Penasa*

ABSTRACT: The aim of this paper is to investigate whether the right to know one's genetic origins (RKGO) encounters significant differences in the level of guarantee when it applies to adoption, to assisted reproduction or to surrogacy. The results of this analysis are aimed at understanding the degree of effectiveness of this right in different legal systems. To this end, the main features of the right to know one's genetic origins are carefully considered, the research being based both on legislative and on jurisdictional materials. Namely, the essay focuses on information and consent, on the structure of relevant regulation in the balance between collection and storage of personal information or protection of anonymity and privacy and, finally, on the most crucial factor for the enforcement of this right, namely time.

KEYWORDS: Right to know genetic origins, anonymous birth, assisted reproduction, surrogacy, personal identity

SUMMARY: 1. The right to know one's genetic origins: theoretical background - 1.1. A right for the future - 1.2. The right to know one's origins beyond state borders - 2. From the European Court of Human Rights to domestic judges: the case of anonymous birth - 2.2. A necessary legislative balance in the French and Italian cases - 2.3. Summing up: the right to know and anonymous birth between past, present and future - 3. Applying the right to know to the field of assisted reproduction and gamete donation - 3.1. Right to know and assisted reproduction: just a matter of regulation? - 4. Right to know and surrogacy: the triplication of motherhood(s) and the best interest(s) of the child - 4.1. Surrogacy and Parliament: right to know, genetic link and social/intended parenthood - 4.2. Surrogacy and courts: self-restraint, the concrete best interest of the child and social parents' duty to disclose - 4.3. Surrogacy and the relevance of genetic and biological ties: a multifaceted issue - 5. Concluding remarks: the right to know one's genetic and biological origins; a relational and multidimensional right.

1. The right to know one's genetic origins: theoretical background

The starting point to address the evolution of the right to know one's genetic origins in the contemporary context of fundamental rights' protection and in the specific area of law and genetics can be found in its theoretical background and its legal acknowledgement in international treaties and in national laws.

We can start from the assumption that the right to know one's genetic origins is a right linked to the "understanding of who we are and how we are connected to others".[1] Therefore, it should be recog-

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ISSN 2284-4503

nised and granted both by national States and international conventions, because it is inherent to individual autonomy and human dignity. In fact, the right to know one's genetic origins gives an individual a freedom connected to personal identity, i.e. the liberty to choose what meaning to assign to the genetic components of individual identity. Acknowledging the right to access information on genetic origins makes the individual free to choose whether to obtain them or not; depriving the individual of such rights means denying a liberty which is strictly connected to the development of personhood.[2] The importance of recognising this right reveals the value of pluralism in contemporary democratic societies: if a legal system regulates access to genetic information, then the State proves to be aware of the inherent diversity of human beings.[3] Not all of us need to know such information but some do and this depends on the different paths we follow to develop our personhood. It is not for the State to deprive individuals of such possibility, especially if it is easily available, but rather it is a matter of individual choice to decide whether to get access to such information or not.

1.1. A right for the future

Above all, the most relevant aspect of the right to know one's genetic origins is that it is a right for the future. In fact, it is granted to children or to a child-to-be (unborn or unconceived) and it could be enforced only once they are adults. This profile makes this right absolutely interesting when dealing with the relationship between scientific development, fundamental rights and individuals' aspirations.

With regard to this feature of the right, it has been noted that, once a State decides to regulate the right to know, for example, for children born through assisted reproduction with gamete donation, it is regulating a possibility that will be effective several years later. This means, for example, that if anonymity in gamete donation is abolished, it is questionable whether the right to know genetic origins should be applied retroactively: the decision to donate may be, in fact, also influenced by the anonymity of donation.[4] Thus, the notion of the right to know one's genetic origins should be carefully considered, as it raises some very important questions and complex issues.

The most significant legal acknowledgment of the right of a person to be able to access information on his/her parental origins is Article 7 of the International Convention on the Rights of the Child (CRC), which provides that the child shall have "as far as possible, the right to know and be cared for by his or her parents". The Convention, moreover, provides that States have an obligation to ensure the effectiveness of these rights "in accordance with their national law and their obligations under the relevant international instruments in this field". On the matter, in 2002, the UN Committee on the Rights of the Child recommended that all States shall take all necessary measures, in relation to

[1] V. RAVITSKY, *Autonomous Choice and the Right to Know One's Genetic Origins*, in *Hastings Center Report*, March-April 2014, 36.

[2] On these matters see K. WADE, *Reconceptualising the Interest in Knowing One's Origins: A Case for Mandatory Disclosure*, in *Medical Law Review*, 28, 4, 2020, 731 ss.

[3] In this perspective see also V. RAVITSKY, *Autonomous Choice and the Right to Know One's Genetic Origins*, cit., 37.

[4] E. FARNÓS AMORÓS, *Donor anonymity, or the right to know one's origins?*, in *Catalan Social Sciences Review*, 5, 2015, 5.

ISSN 2284-4503

the superior interest of the minor, to allow children "to obtain information on the identity of their parents, to the extent possible".[5]

The CRC creates a strong link between the right of the child to have information on his/her parents and the empowerment of the child, which represents the core of the whole convention: children are considered as persons, therefore they have always the right to obtain information on their lives, situation and identity and they also must be considered (and their opinion heard) in any decision concerning them.[6] Therefore, from the viewpoint of the child, the right to know assumes a very crucial role for the development of individual personality.

We should, nevertheless, bear in mind that we are not dealing with an absolute right: the right to know one's genetic origins (RKGO) is instead a relational right. The existence of the possibility to claim the acknowledgment of this right means, per se, that other people are involved in such a request. Indeed, a person's genetic origins are necessarily linked to the "source" of this information, namely a parent or an ancestor. For this reason, the RKGO could be considered as a relational right and involves a complex balance between the right to know, on the one hand, and the right to privacy, on the other hand. As we will see in the following paragraphs, though, these are not the only interests involved and the matter is even more complicated.

This assumption has two implications. The first one is that the RKGO is crucial to the development of personal identity. Questions related to ancestral origins and to the human basic need to know one's provenance are essential parts of the building of personal identity and self-awareness.[7]

The second consequence is quite challenging from a legal point of view, because it involves legal regulation, legislative approach towards new reproductive technologies and family types, but also individual stories, their claim in courts and consequent judicial decisions. If the right to know one's genetic origins is a relational right, then its level of guarantee and enforcement, as well as the role of "others" (i.e. the biological mother, the gamete donor, etc.) depends on the legal regulation or on court decisions. Both legislative enforcement and court adjudication, though, depend on the theoretical construction of this right (a freedom connected to the development of personhood) and on its entrenchment in the constitutional tissue of the legal order concerned, either through a connection in the constitutional text or indirectly by means of international ties.

In all contexts, anyway, the actual degree of acknowledgement is linked to the interpretation given, from time to time and case by case, to the essence of such a right. For example, it has to be considered that there are several and very relevant differences between the legal solutions available. As we will see in the final paragraph, they depend on the degree of information that could be accessed by the child: RKGO is an "umbrella term" that covers the "medical aspect" (i.e. access to information on

[5] Concluding observations, recommendations 31 and 32, CRC/C/15/add.188, 8). See E. FARNÓS AMORÓS, *Donor anonymity, or the right to know one's origins?*, cit., 5.

[6] On the Convention see J. TOBIN (ed.), *The UN Convention on the Rights of the Child: A Commentary*, Oxford, 2019.

[7] The literature in favour and opposing this assumption is very rich. For some references see V. RAVITSKY, *Donor Conception and Lack of Access to Genetic Heritage*, in *American Journal of Bioethics*, 16, 12, 2016, 45-46; S. GOLOMBOK, *Disclosure and donor-conceived children*, in *Human Reproduction*, 32, 7, 2017, 1532-1536.; G. PENNINGS, *Disclosure of donor conception, age of disclosure and the well-being of donor offspring*, in *Human Reproduction*, 32, 5, 2017, 969-973.

health, medical history or relevant genetic information of the parent/donor), the "identity aspect" (i.e. information on the biographical history of the parent/donor, without revealing his/her full identity) and the "relational aspect", which means access to full identity to have the possibility to establish a connection.[8]

Moreover, the degree of enforcement depends on the weight of this right in the balance with other fundamental rights. Therefore, as we will discuss more in detail in the next paragraphs, the legal enforcement and effectiveness of the right to know one's genetic origins depend on the instrument of its regulation at a national level. On the other hand, though, its regulation and, possibly, its jurisdictional acknowledgment depend on the theoretical background on which this right is founded.

1.2. The right to know one's origins beyond State borders

Another relevant aspect of this right is that it often involves cross-border issues. Indeed, it is not infrequent that the genetic background of a person who wants to take advantage of the right to know raises cross-border issues. It might happen with adoption, but also with medically assisted reproduction and with surrogacy. Intending parents might go abroad to adopt a child, or to realise their desire to become parents through reproduction technologies.

In these cases, the effective enforceability of this right depends not only on its regulation at a national level, but also on its acknowledgement in the State in which genetic origins might be found or on the degree of reciprocity of the two legislations. In some cases, the combination of two different regulations might cause a clash or, more easily, might reveal a lack of effectiveness.

For this reason, the analysis of the RKGO should move from a focus on its main features, which encounter different levels of regulation not only depending on the legal system involved, but also on the specific situation in which it has to be applied.

The aim of this paper is therefore to investigate whether the RKGO encounters significant variations when it applies to adoption, to assisted reproduction or to surrogacy. The results will allow us to understand its degree of effectiveness in different legal systems and the need to identify a minimum standard to grant its applicability notwithstanding the intrinsic differences between national regulations. To this end, the main features of the right to know genetic origins will be considered. Namely, the essay will focus on information and consent, on the structure of relevant regulation in the balance between collection and storage of personal information or protection of anonymity and privacy and, finally, on the most crucial factor for the enforcement of this right, which is time.

2. From the European Court of Human Rights to domestic judges: the case of anonymous birth

Adoption is the first area in which RKGO has been recognised and regulated. For example, in the UK adopted children have the right to see their original birth certificate once they reach the age of 18

[8] V. RAVITSKY, *The right to know one's genetic origins and cross-border medically assisted reproduction*, in *Israel Journal of Health Policy and research*, 6, 3, 2017, 2.

ISSN 2284-4503

(16 in Scotland).[9] In Italy, the law on adoption was amended in 2001 to provide for the right of the child to access information on his/her biological parents once he/she is 25 years old.[10] In Spain it is now provided by Law no. 54 of 2007 and it is possible once the adopted child reaches the age of 18.[11]
In the broad field of adoption, the discipline of anonymous birth in recent years has raised some interesting legal issues concerning the content of the right to know genetic origins.
Interestingly enough, similar issues emerged in Italy and in France and were decided by the European Court of Human Rights. In Italy, a significant number of cases concerning people adopted after anonymous birth who applied for disclosure of information on their origins ended up before courts because of the impossibility to get in contact with the biological mother, her refusal or her death. As we will see *infra*, the balancing between the offspring's fundamental right to know their origins, on the one hand, and the mother's right to privacy, on the other hand, might bring to different solutions, especially when the relevant legal framework on the matter is not clear.
Actually, in this respect, it is worth pointing out that the case law of the ECHR on the RKGO always concerns the interpretation of the right to private and family life and, in this context, is considered a matter of personal identity. Most of the case law of the ECtHR developed in the last decades concerns cases of adoption or anonymous birth, whereas there has not yet been case law on the RKGO after assisted reproduction.[12]

2.1. Adoption, anonymity and the European Court of Human Rights

In particular, the starting point for any investigation over the nature of this right is the case of *Gaskin v. UK*, decided in 1989. Here, the applicant claimed that the refusal to access to his personal and confidential information on the part of the City Council violated Article 8 (right to a private life) by failing to meet its positive obligation to give him access to the requested information. After his mother's death, Mr. Gaskin had been taken into the care of the local City Council and had stayed with foster parents until he was 18 years old. He then asked for discovery of his case records, including information on his family of origin and the administration refused on grounds of public interest. The case went to the European Court of Human Rights, which found a breach of Article 8.
In the opinion of Strasbourg judges, a person who has been given in custody during childhood has a "vital interest, protected by the Convention, in receiving the information necessary to know and to understand their childhood and early development".[13] On the other hand, the Court noticed that British law, by making access to records dependent on the consent of the contributor, can abstractly be considered compatible with Article 8 ECHR. Nevertheless, if it is within the margin of appreciation that a state can decide to subject access to records to third parties' consent, then it should in any

[9] This has been possible since 1975 and is now regulated by sections 60 ff. of the Adoption and Children Act 2002. It was previously provided by the Children Act 1989. For further information see https://www.gov.uk/adoption-records
[10] See Article 28 of Italian Law no. 184 of 1983, on adoption.
[11] Article 12 of the Spanish law on international adoption (ley 54 of 2007). See E. FARNÓS AMORÓS, *Donor anonymity, or the right to know one's origins?,* cit., 4.
[12] European Court of Human Rights, *Gaskin v. the United Kingdom*, appl. n. 10454/83, 07/07/1989, *Odievre v. France*, appl. n. 42326/98, 13/02/2033; *Godelli v. Italy*, appl. n. 33783/09, 25/09/2012.
[13] *Gaskin v. UK*, para 49.

ISSN 2284-4503

case provide for an exception for cases in which the person who should consent is not available or improperly refuses disclosure.

The Court finally stated that the principle of proportionality is respected if the national regulation provides for an independent authority's decision when a contributor fails to answer or withholds consent. The lack of such procedure was found to be in breach of the claimant's right to personal and family life.

Gaskin represents a very significant precedent in the case law of the ECtHR with regards to the right to know personal origins, for two reasons. Firstly, the Court defined its nature, making it a "vital interest" and connecting the right to the development of the individual. Secondly, the Court started to outline procedural rights that contribute to grant effectiveness to the substantial right to know one's origins. Indeed, the need that an independent authority could evaluate the individual request to have access to personal records, in the event of the other parties involved improperly refusing to give consent, may be the only practicable solution to realise an appropriate balance of fundamental rights.[14]

The following step on this path is represented by the Grand Chamber's decision in *Odievre v. France*, in 2003. Here, the Court excluded the violation of Art. 8 ECHR by French authorities, but the case represents a very important precedent because it was the first time in which the Court dealt with the issue of anonymous birth. Indeed, the Court confirmed that the right to know has to be traced back to the provisions on private and family life, because it is a matter of relevance to personal development, which is protected by Article 8 ECHR. In particular, the Court noted that birth and the circumstances of birth form a part of the identity, and the right to respect for private life, of the adult.[15]

Nevertheless, the Court excluded the breach of this right in the case in question. Indeed, the Court traced a balancing between the claimant's "vital interest in its personal development" and "a woman's interest in remaining anonymous in order to protect her health by giving birth in appropriate medical conditions" (para 44). By taking into account the specificity of the French law, the general interest in protecting the life and health of women giving birth and children and the fact that the claimant was given access to non-identifying information about her mother and natural family, the Court excluded the violation of Article 8 ECHR. It should be noted that, for the purposes of the Convention, the Strasbourg judges underlined that the right to private and family life was protected by providing the claimant with the possibility to know some information on her roots, safeguarding at the same time the interests of third parties (para. 48). Finally, in the opinion of the Court, States have a margin of appreciation in the determination of the means through which they achieve the aim of reconciling those competing interests. In the case in question, the French legislation respects this margin of appreciation and therefore does not violate the right to respect for private life of the claimant.[16]

[14] Other decisions concerning the RKGO have been issued with reference to paternity testing. See *Mikulic v. Croatia*, appl. n. 53176/99, 7/02/2002; *Jäggi v. Switzerland*, appl. n. 58757/00, 13/07/2006; *Backlund v. Finland*, appl. n. 36498/05, 6/07/2010.

[15] *Odievre v. France*, para 29.

[16] E. STEINER, *Desperately Seeking Mother – Anonymous Births in the European Court of Human*, in *Child and Family Law Quarterly*, 15, 4, 2003, 425-448.

The use of the margin of appreciation doctrine by the Court, in circumstances in which a difficult balancing of interests has to be made by the state legislator, can lead us to some reflections. It is not unusual, for the Strasbourg judges, to refer to these criteria when the balancing of two competing rights under the Convention is so difficult to be drawn and when this assessment recalls several aspects of the national legal framework (such as, in this case, the prevention of abortions and of illegal abandonment of children and the protection of vulnerable subjects).[17]

Interestingly enough, the French Conseil Constitutionnel was involved in a preliminary question of constitutionality on the same matter, ten years after the ECtHR decision.[18] Once again, the law was found to be compatible with the Constitution. In this circumstance, the Conseil considered that the identity of the mother was correctly protected by the French law, which provides that the woman can object to disclosure of her identity even after her death. At the same time, the right to know the origins of the child are protected because there is the possibility to apply for the disclosure of the woman's identity and the mother must be informed, at the moment of birth, that she could consent to reveal her data, once the child is an adult. In the opinion of the judges, the right of the child is guaranteed "as far as possible" and this is considered to be enough with respect to the general aim of the law on anonymous birth, which is intended to avoid the dramatic events we have referred to above. A critical aspect of this decision, which is also underlined in the commentary that the Council offers together with the decision, is that the individual's right to know his/her origins does not have autonomous identity. Indeed, it is completely dependent on the right of the mother to choose not to be named in the birth certificate and is not at all considered as an autonomous right. Actually, it should be pointed out that the Conseil Constitutionnel refers to "interests" of the woman and of the adult child which must be assessed by the law-maker.[19] The deference that the Conseil shows for legislation reveals the difficulties in tracing a line in the definition of the two competing legal positions.

2.2. A necessary legislative balance in the French and Italian cases

It was in the same years that a similar case, concerning the Italian provision on anonymous birth, ended up before the European Court of Human Rights and, a few months later, also before the Constitutional Court.[20] Here, the results were pretty different from the French ones, as the ECtHR found a violation of Article 8, whereas the Constitutional Court found a violation of Articles 2 and 3 of the Italian Constitution.

[17] The margin of appreciation doctrine was also used by the ECtHR in the leading abortion case *A, B, and C v. Ireland*, appl. no. 25579/05, 16/12/2010. In comment to this decision and on the use of the margin of appreciation see S. McGuinness, *A, B, and C leads to D (for Delegation!)*, in *Medical Law Review*, 19, 3, 2011, 476-491; J.N. Erdman, *Procedural abortion rights: Ireland and the European Court of Human Rights*, in *Reproductive Health Matters*, 44, 2014, 22-30.

[18] French Conseil Constitutionnel, Decision no. 2012-248 QPC, 16/05/2012.

[19] Literally: "l'équilibre ainsi défini entre les intérêts de la mère de naissance et ceux de l'enfant", Decision no. 2012-248 QPC, para. 7.

[20] The ECtHR decision is *Godelli v. Italy*, appl. n. 33783/09, decided on 25 September 2012; the Constitutional Court decision is no. 278/2013.

Under Italian law, a woman who gives birth has the right not to be named in the child's birth certificate.[21] In this case, the law provides for the mother's right to remain anonymous; the child is therefore given up for adoption and the relevant law on adoption is applicable with regard to the creation of a legal link between the child and the adoptive parents. The legal issues before both the Strasbourg Court and the Italian Constitutional judge concerned the unreasonableness of the difference between the right to know the child's biological origins, provided by the law on adoption,[22] and the exclusion of this possibility in the case of anonymous birth.

In this respect, we should firstly underline that, in *Godelli v. Italy*, in 2012, the Strasbourg Court recalled the precedent represented by *Odievre v. France*, reconfirming the applicability of Article 8 ECHR, because access to information about one's origins and the identity of one's natural parents is an important part of the development of personhood, protected by the right to respect for private and family life.[23] Moreover, and differently from the previous case, Italian law provides no mechanisms to balance the interest to anonymity of the mother and those of the child, once an adult, to know his/her origins. Therefore, the complete lack of any possibility to request the disclosure of the information concerning the natural mother (not even non-identifying information) is a decision that oversteps the margin of appreciation. In principle, States can choose between the several possibilities available in order to protect both interests, but the complete sacrifice of the position of the child amounts to a violation of the Convention. Unlike French law, the Italian legislation does not allow the woman to change her mind at a later stage and decide to identify herself.[24]

As in the French case, though, the position of the ECtHR is very much focused on parental rights, rather than on the essence of the right to know genetic origins. Indeed, the breach of the Convention and also of the margin of appreciation was found, in *Godelli*, in the absence of the possibility *for the woman* to change her mind, and not in the disproportionate sacrifice for the rights of the child.[25] It is no coincidence that neither of these judgements makes reference to the UN Convention on the Rights of the Child, which expressly recognises and promotes the right to know, as far as possible, one's origins.

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[21] DPR 396/2000, Article. 30. It is worth noticing, however, that this possibility is explicitly excluded in case of artificial reproductive technologies, as provided by Article 9, par. 2, of law no. 40/2004.

[22] The Italian law on adoption, Law n. 184/1983, provides for the right of the child to have access to the information concerning his/her biological origins and the identity of his/her biological parents at the age of 25. To this end, he/she should file an application before the competent juvenile court.

[23] The case brought before the European Court of Human Rights concerned a woman, born from an anonymous birth, who filed an application to have access to the information concerning her biological mother. The Tribunal, applying the law, refused her request; therefore, she appealed to the ECtHR, affirming that the Tribunal's denial and the Italian legal framework violate her right to respect of private and family life, protected by Article 8 of the Convention. The Strasbourg Court acknowledged that the relevant legal framework represented the result of a wrong balancing made by the Italian law-maker between competing fundamental rights and therefore Italy was condemned for violation of art. 8 ECHR. For a comment to *Godelli v. Italy*, see C. SIMMONDS, *An Unbalanced Scale: Anonymous Birth and the European Court of Human Rights*, in *The Cambridge Law Journal*, 72, 2, 2013, 263-266.

[24] On the matter see also A. MARGARIA, *Anonymous Birth: Expanding the Terms of Debate*, in *International Journal of Children's Rights*, 22, 3, 2014, 552-580.

[25] In this respect, see also C. SIMMONDS, *An Unbalanced Scale: Anonymous Birth and the European Court of Human Rights*, cit., 265.

It was a few months later that the Italian Constitutional Court was called to evaluate the compatibility with Constitutional provisions of the law on anonymous birth, and confirmed the principles established in *Godelli*. The Court overruled its precedent decision no. 425 of 2005 (in the Court's words, a 'fully analogous case' in which the question of unconstitutionality was judged as clearly unfounded). In particular, in decision no. 278 of 2013, the Court noted that the irreversibility of the mother's anonymity is unreasonable, as the right of the mother must be balanced with the right of the child to know his/her biological origins, which is protected by article 2 of the Italian Constitution. Moreover, the Court also found a violation of the principle of equality (Article 3 Const.), as this right is granted to adopted children, with the only exception of those born from anonymous birth.

The Court finally suggested that it is for the law-maker to set a balanced system of rules permitting the assessment of the subsistence of the willingness of the mother to remain unknown. The intent of the law, anyway, should be to try to reconcile these two opposite positions, by giving to the child the possibility of accessing information on the identity of the mother.[26]

The Italian Court balanced the two opposite interests. On the one hand, anonymity is justified by the need to protect not only the privacy of the mother, but also her health, and the child's health to avoid risks for the newly born life, and to ensure a framework for the birth to occur in the best possible conditions. On the other hand, consideration is also given to the right of the child, which "represents a significant element within the constitutional system ensuring protection for the person" and which "constitutes one of the aspects of the personality that can condition the intimacy and the very social life of a person as such".[27] It should be underlined, however, that although the irreversible secrecy of the mother's identity and information was found to be unconstitutional, the Court stressed the need for a legal intervention to properly address the necessity to reconcile these two opposite positions. Actually, this is a problem which is shared among most jurisdictions: the provision on anonymous birth is made to safeguard the safety of birth and to avoid or reduce abortion. Nevertheless, this interest in protecting the vulnerability of a particular situation cannot completely cancel the right of a person to obtain information on his/her biological origins, especially several years after birth.[28]

In Italy, at the moment, no legislative intervention has followed the mentioned decisions; therefore, every time a child whose mother did not want to be named on the birth certificate wants to have access to the information on his/her origins, he/she has to go to court. Courts gave very different interpretations of the principle of disclosure, until a landmark decision by the Court of Cassation in 2017.[29] Here, the Italian Supreme Court stated that the decision by the Constitutional Court clearly indicates

[26] For a comment to the decision of the Italian Constitutional Court, see V. COLCELLI, *Anonymous Birth, Birth Registration and the Child's Right to Know Their Origins in the Italian Legal System: a Short Comment*, in *Journal of Civil and Legal Sciences*, 1, 2012, 101.

[27] Italian Constitutional Court, decision n. 278 of 2013, para. 4. Translation by the Constitutional Court, available at https://www.cortecostituzionale.it/documenti/download/doc/recent_judgments/278-2013.pdf

[28] Several states, in this respect, provide that the woman must leave non-identifying information that the child could obtain once he/she reaches a sound age. See C. SIMMONDS, *An Unbalanced Scale: Anonymous Birth and the European Court of Human Rights*, cit., 265, making reference to Austria and Germany.

[29] Court of Cassation, decision no. 1946 of 2017. On previous decisions by Italian tribunals see the following address https://www.biodiritto.org/Biolaw-pedia/Giurisprudenza/Tribunale-di-Milano-sent.-11475-2015-parto-anonimo and the essay by S. AGOSTA in this volume.

a principle that judges can (and shall) follow in order to recognise the right to know the origins of a child born from anonymous birth. Even if the Constitutional judges said that it is for the law-maker to identify the most appropriate point of balance between the opposite interests of the mother and of the child (once an adult), in the absence of new discipline on the matter, judges should make all reasonable efforts to ascertain the present will of the mother and to ask her consent to disclose her identity. Some time later, the Court of Cassation also specified that, in case the woman is already dead at the moment of such request, then her privacy can never prevail over the right of the son/daughter to have access to information on his/her origin.[30]

2.3. Summing up: the right to know and anonymous birth between past, present and future

Beyond the concrete national disciplines on this specific matter, in this context it is worth stressing the delicate role of the Strasbourg Court in addressing the main features of the right to know one's origins in this field. In matters concerning rights connected to personal identity and complex balancing, the ECtHR often plays a decisive role, which proves to influence subsequent national decisions. In ethically controversial matters, Strasbourg decisions could concretely help in drafting a minimum standard for the protection of fundamental rights in Europe and contribute to an advancement of the legal debate on the matter.[31]

The discipline of anonymous birth raises very complicated questions, because it requires a careful balancing between opposite positions which deserve due protection, being linked to fundamental rights of the person. This issue reveals several aspects of interest that help us to take a further step in the identification of the basic features of the right to know one's origins.

First of all, when born from an anonymous birth, the right to know must *always* be balanced with the right to privacy of the mother. Interestingly enough, the reason to protect the mother's identity is also rooted in the need to protect the child: the discipline of anonymous birth, in fact, is created to allow the woman to give birth in a protected and safe environment, also in order to avoid a decision on her part to give birth in risky situations or to have an abortion. Therefore, the right to know must be balanced also with the right of the child to have a safe birth. It is not only a matter of conflict between the rights of the mother and those of the child. As previously outlined, the right to know has a profound relational nature, because it can never be considered an absolute right, but it always has to be considered together with other fundamental rights.

Secondly, this right has a very particular time frame. Indeed, the concrete situation in which the relevant legal position arises occurs in a given time and context, which is the time of birth of the child and the will of the mother not to be named in the birth certificate. Nonetheless, the right itself can be enforced only several years after the moment when the relevant circumstance occurred, which is the moment in which the child becomes an adult or reaches the age established by law to apply to have access to information on his/her origin. As we will see in the following paragraphs, this is a feature of the right that is relevant also in the other fields of its application. In this specific context, time represents the factor on which the balancing is based. The need to ascertain the current will of the

[30] Court of Cassation, ordinance no. 3004 of 2018.

[31] See D. FENWICK, *Abortion jurisprudence' at Strasbourg: Deferential, avoidant and normatively neutral?*, in *Legal Studies*, 34, 2, 2014, 214-241.

woman depends on the time that has elapsed, which is the criterion that permits disclosure of the identity of the mother, which otherwise would remain secret in order to protect the vulnerability and the need for safety mentioned before. This is the main reason that brought both the Strasbourg Court and the Italian Constitutional judge to hold that the Italian law which does not provide for instruments to ascertain the current will of the mother is disproportionate and unconstitutional. Asking the mother should better satisfy this controversial balance.

In brief, the need to protect a potentially vulnerable woman and her decision (in the present) to give birth to a child without being named in the birth certificate, could be determinant and could – with adequate guarantees – even be prevalent over the right of the "future" child (once an adult) to have effective access to the information concerning the woman's identity. Exceptions should also be made in the event of the woman being already dead when the child, having reached adulthood, wants to have access to information on his/her origin.

Thirdly and finally, in the field of adoption and in the specific context of anonymous birth, genetic information has very little relevance. Indeed, we have rather been referring to a right to know origins or information on the natural mother. In this field, genetic information *per se* is not at the centre of the legal guarantee of the right, which is rooted instead in the inherent need for a person to know the circumstances of his/her birth as a matter of personal identity (i.e. identity and relational aspects, in conformity with the distinctions we have drawn before). Actually, in this context, genetic information is linked to the disclosure of the medical data of the mother, which should in any case be made accessible for health reasons. Indeed, in all cases health proves to be prevalent over the need to respect or protect the privacy of the natural mother.

3. Applying the right to know to the field of assisted reproduction and gamete donation

The features and the understanding of the right to know reveals different facets if we move from anonymous birth to medically assisted reproduction.

First of all, when dealing with assisted reproduction with gamete donation, we are considering voluntary donation, which means that the donor consciously offers his/her genetic material to contribute to the creation of new human life. Differently, the discipline on adoption and anonymous birth is based on the need to protect an existing child who cannot be raised by his/her natural parents. Moreover, and differently from anonymous birth, the child is not yet born, nor is he/she even conceived, at the moment of donation, that is when is it necessary to inform the donor on the future possibility of disclosure. Hence, the discipline concerning the right to have access to information on biological/genetic origins foreruns both conception and birth and is intended to determine the concrete fact (birth) that causes the applicability (in a quite remote future) of the relevant legal discipline at the time of donation.

Therefore, there is no balance to be struck between the interests of the donors and the right to know the genetic origins of the child. Yet, the regulation of gamete donation and the continual technologi-

cal progress[32] in this field have caused a significant scientific debate that counterposes those in favour of donor disclosure to those that prefer anonymity of donation.

In this particular area, the starting point for a legal dissertation on the inherent nature of the RKGO is that it is a widely common opinion that the disclosure of conception is strongly recommended both in medically assisted reproduction in general and with gamete donation.[33] The possibility to have access to information on conception and birth is regarded as a very important part in the construction of a child's identity.[34] Nevertheless, anonymous donation is still quite widespread at a normative level and, in any case, donor records are not necessarily kept for a long time.[35] It is worth mentioning, moreover, that in some countries where donor disclosure has been regulated for a long time, there are some studies which show that intending parents have a reluctant attitude towards the disclosure of the circumstances of conception.[36] In other words, parents who have access to assisted reproduction with gamete donation may choose not to disclose to their offspring that there was a third party contribution to their existence, with the fear of possible negative or disruptive consequences for their family equilibrium. It is not surprising that this problem is relevant almost only among heterosexual couples, as same-sex parents or singles are forced to give such explanations to their children at a certain stage and, therefore, the path towards the discovery of genetic origins is more natural.[37]

Notwithstanding this wide agreement on the need to inform the child that he/she was conceived and born thanks to reproduction technologies, the possibility to have access to information on the donor's identity is more controversial.[38] Therefore, the right of a child to have access to information on his/her origin is abstractly guaranteed by both approaches, but in the first case it is limited to the information on the circumstances of conception and does not extend to the identity of the donor. In particular, it has to be remarked that, in any case, this right is hardly enforceable: indeed, even if the law provides for the opportunity to inform the child on the method of conception, obviously it is not

[32] See, for example, the possibilities offered by mitochondrial donation, which has already been regulated in the UK, or the application of gene editing (CRISPR-Cas9) to embryos.

[33] See for example: ASRM (Ethics Committee of the American Society for Reproductive Medicine), Informing offspring of their conception by gamete or embryo donation: a committee opinion, in Fertility and Sterility, 100, 2013, 45–9.

[34] For example, V. RAVITSKY, *The right to know one's genetic origins*, cit., is very much in favour of disclosure. More recently, on the European perspective on this issue see K. WADE, *Reconceptualising the Interest in Knowing One's Origins: A Case for Mandatory Disclosure*, cit.

[35] V. RAVITSKY, *The right to know one's genetic origins*, cit., 2.

[36] A. BREWAEYS et al., *Anonymous or Identity-Registered Sperm Donors? A Study of Dutch Recipients' Choices*, in *Human Reproduction*, 20, 2004, 820, finding that only 17% of parents choosing an anonymous donor intended to disclose to the child the circumstances of his or her conception; C. GOTTLIEB et al., *Disclosure of Donor Insemination to the Child: The Impact of Swedish Legislation on Couples' Attitudes*, in *Human Reproduction*, 15, 2000, 2052; M. KIRKMAN, *Parents' Contributions to the Narrative Identity of Offspring of Donor-Assisted Conception*, in *Social Science & Medicine*, 57, 2003, 2234–35. See also V. RAVITSKY, *The right to know one's genetic origins*, cit., 3.

[37] E. FARNÓS AMORÓS, *Donor anonymity, or the right to know one's origins?,* cit., 7.

[38] A strong opposition is expressed by I. DE MELO MARTÍN, *The ethics of anonymous gamete donation: Is there a right to know one's genetic origins?*, in *Hastings Center Report*, 44, 2, 2014, 28-35.

possible to force parents to do so,[39] unless the application of medically assisted reproduction technologies or donated gametes is written in the birth certificate.[40]

The problem is that, in any case, the right of the child to have access to information on the donor's identity is not enforceable, unless the relevant legislation provides for concrete instruments to make it effective. More specifically, if the law does not provide for the duty, firstly, to tell the child about assisted reproduction, and, secondly, to disclose the donor's identity at the request of the child (once he/she is an adult), then it is not possible to obtain information on the personal identity of the donors, except to raise a question of constitutional legitimacy. The only exception, obviously, regards information that is necessary for medical reasons. In this regard, several solutions on the instruments of legal regulation have been proposed. They space from the provision of a sort of notification to be sent by letter when the child is of sound age (for example, at 16 or 18 years old) to a set of socio-cultural incentives to make parents comfortable with the idea that telling the truth to the child will not be disruptive for their family relationships.[41]

Another relevant aspect concerns original intentions.

Whereas in anonymous birth we can identify a significant connection between the child and the biological mother, which regards the carrying of the pregnancy and the decision to give the child up for adoption, in gamete donation the biographical link is much too weak to offer a contribution to the development of the personality of the child. In fact, when dealing with assisted reproduction (with or without donation), the central role is played by the intending parents, whose extremely strong desire to have a child is the key determinant of the new life. In assisted reproduction, the genetic material comes from intending donors, who consciously offer their gametes to allow other people to become parents. They are aware of their role since the beginning and they know that there will be no further relationship with the child. Similarly, the parents are aware they are benefiting from the gametes of an unknown donor to realise their desire to have a family.

In these circumstances, does a full understanding of the right to know one's origins really contribute to the development of individual personality, in the sense we have been dealing with in the field of anonymous birth? Here, we do not have an individual biographical story to discover. Relevance has to be given to the stories of intending parent(s), but the donor seems to have a more marginal role.

Therefore, if we take this perspective, then access to genetic information might be even more important than in the previous example. Indeed, in this case, genetic information is the data that a person may be really interested in, for their medical relevance.

[39] On this, S. GOLOMBOK et al., *The European study of assisted reproduction families: The transition to adolescence*, in *Human Reproduction*, 17, 3, 2002, 830-840, makes reference to the uncertainty of leaving the decision to parents.

[40] K. WADE, *Reconceptualising the Interest in Knowing One's Origins: A Case for Mandatory Disclosure*, cit., 749.

[41] The topic has been widely investigated. Beyond K. WADE, *Reconceptualising the Interest in Knowing One's Origins*, cit., see M. GARRIGA GORINA, *El conocimiento de los orígenes genéticos de la filiación por reproducción asistida con gametos donados por un tercero*, in *Derecho Privado y Constitución*, 21, 2007, 167-228; Nuffield Council on Bioethics, *Donor Conception: Ethical Aspects of Information Sharing*, London, 2013.

Among European countries, Sweden became the first one, in 1985,[42] to legally regulate gamete donation and to recognise the right for all offspring "to obtain identifying information about the donor when they are sufficiently mature".[43] For the law to be properly effective, at least two conditions must be fulfilled: first of all, recipient parents should tell their children about the way they were conceived; secondly, the offspring should be made aware of this possibility and should apply to the competent authorities for donor data disclosure.[44] Quite interestingly, several arguments can be made either to favour or to oppose donor identity disclosure to the offspring. In any event, it is a matter for national regulation and might deeply differ on a State by State basis, given also the specificities of the discipline on medically assisted reproduction.

3.1. Right to know and assisted reproduction: just a matter of regulation?

As we have seen, from a biographical or anthropological viewpoint, the need to discover one's origins might have different dimensions if it is related to anonymous birth or to assisted reproduction. In the former case, the relevance of the disclosure is represented by the identity of the natural mother and by her story; in the latter case, the right of the child is more closely linked to the need to know his/her genetic asset. The need to know the identity of the donor is somewhat rare and the possibility to have access hardly takes prevalence over the donor's right to anonymity and privacy.

The ways in which a person born through gamete donation can have access to information on his/her donors is strictly regulated by legislation. In case of lack of a dedicated discipline, there is no enforceability for the right to have access to information on the donor, with the only exception of medical data. In general terms, the regulation of medically assisted reproduction frequently adopts the approach of donor anonymity, with the exception of the disclosure of health data. Therefore, the most widespread model seems to be the one concerning access to the medical aspect, and less frequently to the identity or even the relational aspects.

Moreover, also in this field, the time factor has a crucial role. Even if international conventions and national laws provide for the right of the child to know his/her origins, which means being informed of the circumstances of conception, the enforceability of the right to know one's identity in the case of gamete donation is enforceable only when the child has grown up. This means that, even if there is a general (moral) obligation for the parents to tell the truth and to explain to the child how he/she was conceived, the person can have access to the information on the donor only once he/she is 18-25 years old (depending on the national legislation) or, before, only for exceptional circumstances (such as health needs). The long time between donation and the possibility of disclosure requires the law to provide for concrete mechanisms for the enforceability of the right, which include the regula-

[42] The law is no. 1140/1985, known as the Genetic Integrity Act, amended in 2006 (2006:351) and available at http://www.smer.se/news/the-genetic-integrity-act-2006351/ .

[43] S. ISAKSSON et al., *Two decades after legislation on identifiable donors in Sweden: are recipient couples ready to be open about using gamete donation?*, in *Human Reproduction*, 26, 4, 2011, 853. See also M. DENNISON, *Revealing Your Sources: The Case for Non-Anonymous Gamete Donation*, in *Journal of Law and Health*, 21, 2008, 8.

[44] Some recent studies investigated the impact of the law in Sweden and its effectiveness, as it seems that not all parents told their children about donation. S. ISAKSSON et al., *Two decades after legislation on identifiable donors in Sweden*, cit.

tion of registries. In other words, once the decision to regulate access to information on the donors' identity is adopted, then it is necessary to provide all forms of contact and consent possible to make the right really effective.

On the donor's side, the full disclosure of his/her identity, after more or less twenty years from when the donation took place, might be disincentivizing for a prospective donor. In this regard, regulatory instruments should also take into account the ongoing problem of gamete shortage, which compels some states to purchase gametes from abroad.

Taking into account all relevant variables, it seems that the most balanced solution, in the field of gamete donation, is to balance the right to have access to information on one's identity with the nature of the donation, with the original intent of both donors and intending parents and with the need to maintain the functionality of donation, mainly through the availability of gametes coming from different donors. Therefore, access to health information as well as to general information on origins shall be ensured in order to satisfy the right to know. This has to be done through adequate registries and informing prospective donors and intending parents of the possibility to disclose this information. The decision to reveal the whole identity (i.e. the relational model) might be considered as a matter of discretionary power, in consideration of the different values involved. The most balanced solution seems that of giving to the child the possibility to choose whether to have access to full or partial information and, respectively, to leave the donor the possibility to disclose just some information or his/her full identity.[45]

4. Right to know and surrogacy: the triplication of motherhood(s) and the best interest(s) of the child

Surrogacy is the third context in which the issue of the right to know one's genetic origin is analysed. Here, some of the suggestions already raised with specific regard to assisted reproduction can be easily referred also to surrogacy, especially if we consider that, in many countries where the latter technique is allowed and disciplined, the donation of an ovum by a woman other than the one who will carry the pregnancy is a condition for the lawfulness of this practice.[46] Therefore, in the case of surrogacy, the level of complexity – in social, ethical and legal terms – related to the specific characteristics of this practice becomes even higher, due to the distinction between genetic and biological motherhood, which can occur at the biological/medical level due the specific techniques implemented (ova donation); and the potential separation between gestational and social motherhood, which represents one of the main challenges posed by the regulation of surrogacy agreement effects (more specifically, by the birth of a child via surrogacy).[47]

[45] K. WADE, *A case for mandatory disclosure*, cit., is very much in favour of full disclosure of the donors' identity.

[46] For a recent comparative study, see R. LA RUSSA, *Le pratiche di maternità surrogata nel mondo: analisi comparatistica tra legislazioni proibizioniste e liberali*, in *Responsabilità Civile e Previdenza*, 2, 2017, 683-716.

[47] S. CECCHINI, *Il divieto di maternità surrogata osservato da una prospettiva costituzionale*, in *BioLaw Journal-Rivista di BioDiritto*, 2, 2109, 335, highlights that the involvement of at least one woman who is not part of the couple and who shares the parental project is capable of splitting motherhood into three different roles: biological, genetic and social mother. The biological mother is the one who carries the pregnancy and gives birth. The genetic mother, on the other hand, is generally anonymous and provides the eggs. Lastly, the social moth-

With regard to the first dimension – the separation between genetic and biological motherhood – the recognition of a right to know the identity or at least have access to specific information referring to the ova donor (e.g., of a medical nature) is usually coherent with the existing regulation of assisted reproduction via gamete donation, except when a surrogacy-tailored exception is provided.[48] Accordingly, if anonymity is the rule in the case of ova donation, it will apply also in the case of surrogacy, as well as when the law allows one to obtain – under specific conditions – certain information about the donor (e.g. related to health condition). With regard to the second dimension – gestational *vis a vis* social motherhood – the right to know the identity of the gestational mother becomes crucial in the perspective of the concrete definition of the best interest of the child born via surrogacy.[49] In this case, *ad hoc* rules are usually set forth by national legislations which regulate this practice. Interestingly enough, even where surrogacy is explicitly forbidden by law – such as in Italy, Spain, and France – the matter of knowing one's genetic origins forms part of the broader issue of the right to an identity of the person born. The analysis will focus on the latter dimension, where the right to know one's genetic origins may serve two purposes: as a fundamental right of the newborn via surrogacy; and as a criterion for the acknowledgment of the *status filiationis*, both directly when surrogacy is allowed and regulated or indirectly, in all those cases where the law prohibits the practice and does not provide any *ad hoc* criteria for status determination in the case of cross-border surrogacy. In the former case, the right to know may be part of the balancing between the public and private rights and interests at stake, which legislature is primarily called to define when regulating surrogacy; in the latter case, it can become relevant in the light of guaranteeing the child's right to personal identity within the broader context of the *status filiationis* determination, in which genetic identity seems to be less relevant than – or at least functional to –the need to establish the most adequate normative framework to protect child's personal identity.

4.1. Surrogacy and the Parliament: the right to know, genetic link and social/intended parenthood

If we adopt the perspective of the legislative approach to surrogacy, then, the effective recognition and protection of RKGO depends mainly on the existing regulation of gamete donation in the broader framework of assisted reproduction.[50] At the same time, the legislative choice to provide for the absolute ban of this technique may also be grounded in the aim of guaranteeing the genetic identity of the child born via surrogacy as a public absolute value which would be – together with other public interests, such as the prevention of women's exploitation and the security of the parental relationship – violated by such practice. Italy is a paradigmatic example of this attitude, even if there is not a direct reference to the need of protecting a child's genetic identity at the legislative level as a reason

er, also known as the commissioning mother, is the one who has expressed, together with her partner, her wish to assume full parental responsibility for the newborn child.

[48] See the Portuguese case below.

[49] I. RIVERA, *La complessa questione della maternità surrogata tra rispetto dell'ordine pubblico e protezione del "best interest of the child": un percorso ermeneutico non sempre coerente*, in *Sociologia del diritto*, 1, 2020, 201-222.

[50] On this issue, see from a broader perspective L. POLI, *Artificial reproductive technologies and the right to the truth about genetic and biographic origins*, in *International Journal of Technology Policy and Law*, 3, 1, 2017, 56-67.

to provide for the absolute ban of surrogacy. With regard to the Italian case, it is worth mentioning that the Constitutional Court explicitly referred to women's vulnerability and dignity as constitutional goods violated by surrogacy; it did not mention the child's right to know or the right to genetic identity.[51] In broader terms, the Italian Constitutional Court – albeit in the different context of assisted reproduction via ova donation – clarified that, in the light of the social and cultural evolution which has been characterising the idea of familial and parental relationships in contemporary societies, the issue of the genetic origin of a child is not an essential prerequisite for the existence of a family.[52] At the same time, the separation between genetic and biological ties, on the one hand, and social ties, on the other, in the light of assessing the determination of a parental linkage, does not automatically eliminate the distinct issue represented by the possible will of the child born via surrogacy to know his or her own genetic or biological origins, intended as an expression of the right to personal identity.

If analysed from the latter perspective, the issue of access to information related to the genetic or biological linkage of the child born via surrogacy, both directly or through the legal parent(s), may find an explicit answer when this practice is regulated at the national level. The Portuguese case is particularly relevant. Article 8 of Law 32/2006 on Medically Assisted Procreation (following the reform introduced by Law 25/2016) allows surrogacy, exclusively on an exceptional and altruistic basis, without remuneration and voluntarily, subject to prior authorisation by an independent body, the National Commission for Medically Assisted Reproduction. Within this legal framework, Article 15 introduces the principle of the anonymity of donors and surrogate mother, with the exception of unspecified cases where information on the identity of the donor may also be obtained for important reasons recognised by a court judgment.[53] In 2019 (Law 48/2019), a new exception was provided, which applies also to surrogacy, according to which "the people born as a result of reproductive procedures through the use of gametes or embryos may obtain, from the competent health services, genetic information concerning them, as well as information on the donor's civil identification, obtained from the National Council for Medically Assisted Procreation, provided that they are over 18 years old".[54] The assessment made by the Portuguese Constitutional Court of the anonymity regime is especially relevant in expressing the complexity that the separation between motherhoods inevi-

[51] Italian Constitutional Court, judgments no. 33 of 2021 and 272 of 2017.

[52] Italian Constitutional Court, Judgment no. 162 of 2014.

[53] Art. 15, paragraph 4. See V.L. RAPOSO, *Rise and fall of surrogacy arrangements in Portugal (in the aftermath of decision n. 465/2019 of the Portuguese Constitutional Court)*, in *BioLaw Journal – Rivista di BioDiritto*, 1, 2020, 10/15. See also the Greek legislation, which adopts the principle of anonymity regarding any donation (in this Journal, T. CHORTARA, S. PENASA, L. BUSATTA, *The best interests of the child born via cross-border surrogacy. A comparison between Greece and Italy*, in *BioLaw Journal-Rivista di BioDiritto*, 1, 2016, 189-210. Art. 8 of the Law n. 3305/2005 on the Application of medically assisted procreation provides that medical information referring to the donor must be kept in an anonymous codified form in the Cryopreservation Bank and in the National Registry of Donors and Receivers.

[54] V.L. RAPOSO, *Rise and fall of surrogacy arrangements in Portugal (in the aftermath of decision n. 465/2019 of the Portuguese Constitutional Court)*, cit., 350, clarifies that "Law 32/2006 never enshrined a pure anonymity regime. From the very beginning, Article 15/4 stated that "information may also be obtained on the identity of the donor for significant reasons recognised by court decision". The exact meaning of the wording "significant reasons" was never clarified, but some have asserted that this clause includes situations of severe emotional distress caused by the lack of knowledge of the child's genetic origins".

tably provokes also in legal and constitutional terms. At first, the Constitutional Court (judgment no. 2009) considered justified the mitigated regime of anonymity set forth by Article 15, as it aims to weigh the right to know one's genetic ancestry, which is an expression of the right to personal identity, with other constitutional values, such as the right to found a family and the right to respect of private and family life. In 2018 (Judgment no. 225), the same Court reached a completely opposite conclusion. It stated that, in the light of the centrality of knowledge of one's origins as a fundamental element in the development of personal identity, the regime of temperate anonymity introduced by Article 15 infringes the essential core of the right to personal identity and the right to the development of the unborn child's personality.[55] According to the Court, an opposite legal regime, which enshrines the possibility of anonymity of donors and surrogate mother exclusively when serious grounds for doing so exist, to be assessed on a case-by-case basis, would be a more acceptable solution (§80). For the sake of analysis, it is worth highlighting that the Court associated the desire to know the identity of the person who carried out the pregnancy – the biological mother – to the claim to know one's genetic origins, by acknowledging that pregnancy must be considered a "differentiating personal experience" and that the surrogate mother may become a relevant reference point for the newborn child's biographical itinerary (§ 79). Thus, the Portuguese trajectory in the regulation of surrogacy clearly shows that it is primarily the Parliament's duty to design the legislation on surrogacy – in the case of an absolute ban as well as of conditional admissibility of the practice – taking into account also the issue of the child's origin, both from a genetic and a biological perspective. It must also be clarified that Parliaments enjoy a broad margin of appreciation in performing this assessment, as emerges for instance from the consolidated case-law of the European Court of Human Rights.[56]

In the context of the valorisation of genetic linkage at the legislative level, the existence of a genetic link between the child born via surrogacy and the intended parents can be prescribed by law as an essential condition for the admissibility of the practice. In this case, it is worth noting that the rationale of the requirement is essentially to guarantee the best interest of the child, on the one hand; and that it directly forms part of the discipline on surrogacy and it does not become relevant only *ex post*, when it is usually enforced as a condition to determine the parental relationship with the intended parents (as happens in Italy and according to the ECtHR case-law),[57] on the other hand. From a comparative perspective, section 294 of the South African Children's Act[58] provides that "no surrogate motherhood agreement is valid unless the conception of the child contemplated in the agree-

[55] *Ivi*, 348.

[56] On States' margin of appreciation in the context of surrogacy law according to the ECtHR's case-law, see recently A. MARGARIA, *Parenthood and Cross-Border Surrogacy: What Is 'New'? The ECtHR's First Advisory Opinion*, in *Medical Law Review*, 28, 2, 2020, 418 ff.

[57] See i.e. the ECtHR, case *Mennesson v. France*, 26 June 2014, n. 65192/11, where the genetic link with at least one parent is identified as the criterion for status determination (§100).

[58] Law no. 38 of 2005. See the 2017 Legal Grounds III: Reproductive and Sexual Rights in Sub-Saharan African Courts, edited by the International Reproductive and Sexual Health Law Program, University of Toronto, Centre for Human Rights, Faculty of Law, University of Pretoria, South Africa e Center for Reproductive Rights, New York, Pretoria University Law Press (PULP), 2017, 106-110 (www.pulp.up.ac.za/legal-compilations/legal-grounds); M. SLABBERT, C. ROODT, South Africa, in K. TRIMMINGS, P. BEAUMONT (eds.), *International Surrogacy Arrangements: Legal Regulation at the International Level*, Hart, 2013, 325-346.

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ment is to be effected by the use of the gametes of both commissioning parents or, if that is not possible due to biological, medical or other valid reasons, the gamete of at least one of the commissioning parents or, where the commissioning parent is a single person, the gamete of that person". This requirement is recurrent also in other legal systems in which specific forms of surrogacy are allowed (i.e. Portugal and the United Kingdom). Notwithstanding, South Africa's case is particularly relevant because the Constitutional Court of that legal order has been called to assess the compatibility of such criterion, which is considered part of the best interest of the child, with other constitutional rights, such as the right to privacy, the right to physical and psychological integrity and the right to health of the intended parents that are unable to contribute with their own gametes to a surrogacy agreement, as well as the principle of equality and dignity. According to the Court, which eventually declared the question groundless, the requirement of a genetic linkage with at least one of the intended parents enacts the principle of the best interest of the child born via surrogacy, which finds formal constitutional grounds.[59] Thus, "The requirement of donor gamete(s) within the context of surrogacy indeed serves a rational purpose (…) of creating a bond between the child and the commissioning parents or parent", which is designed "to protect the best interests of the child-to-be born so that the child has a genetic link with its parent(s)" (§ 286).

4.2. Surrogacy and the courts: self-restraint, the concrete best interest of the child and social parents' duty to disclose

If we refer to the link between the RKGO and the determination of the *status filiationis*, the courts' perspective becomes particularly relevant in order to try to understand which normative function genetic ties can play in this specific context, which is independent of the legislative model chosen by a legal order to regulate surrogacy. From a comparative perspective, courts may assume different stances when assessing the role played by genetic origins in the context of determining parental relationships between all the subjects involved in surrogacy agreements.[60]

Courts may be deferential to the choices made by legislature. Accordingly, genetic or biological truth may be considered prevalent even when clear and explicit consent has been given by the intended parents, if an *ad-hoc* rule for determining parenthood in the context of surrogacy has not been explicitly set forth at the legislative level. Within this judicial approach, a judgment of the Supreme Court of Ireland, related to the determination of the parental relationship of a child born via surrogacy, is particularly significant. The High Court had previously ordered the registration in the civil status registry of the intended mother, as the genetic mother of the child, instead of the woman who had given birth to him, on the basis of the existence of both a genetic link (the intended mother's ova had been used) and the parental will. On the contrary, the Supreme Court overturned the High Court's judgment, on the grounds that a legislation on surrogacy and its effect in terms of parenthood does not exist in the Irish legal system and that "It is, thus, quintessentially a matter for the Oireachtas

[59] Article 28 of South African Constitution, according to which "A child's best interests are of paramount importance in every matter concerning the child".

[60] See D. ROSANI, *"The best interest of the parents". La maternità surrogata in Europa tra interessi del bambino, Corti supreme e silenzio dei legislatori*, in *BioLaw Journal - Rivista di BioDiritto*, 1, 2017, 24 ff.

[the Irish Parliament]".[61] The Supreme Court does not assess the merit of the issue, nor does it clarify the relevance of the existence of a genetic link with the intended mother; it merely refers to the appreciation of the legislature, based on the fact that the issues raised – related to the status and rights of children and family – are "important, complex and social, which are matters of public policy for the Oireachtas" and cannot be addressed by a court.[62]

Courts generally recognise the prevalence of the need to guarantee the best interest of the child, which is a very broad concept consisting of a bundle of different rights and principles, where the right to know genetic origin represents only one of the possible relevant components. Therefore, the need to protect one's genetic identity shall not act as a limit for the effective protection of the broader personal identity of the child born via surrogacy, of which genetic or biological linkage is only one of the constitutive elements. In order to show different approaches to the definition of the connection between the best interest of the child and the existence of a genetic link with the intentional parents, it is worth recalling a judgment of the Italian Constitutional Court, related to the disavowal of paternity following a surrogacy agreement (Judgment no. 272 of 2017).[63] According to the Italian Court, "whilst it is necessary to acknowledge a marked preference expressed by the legal order that the status of an individual should reflect the actual circumstances of his or her procreation, it cannot be asserted that the establishment of the biological and genetic parentage of an individual is a value of absolute constitutional significance, as such immune to any balancing operation". Therefore, the right to know or the right to genetic (or biological) identity can be functional to the effective protection of the best interest of the child born via surrogacy. At the same time, genetic identity cannot be intended as an absolute constitutional value, the protection of which would in practice be contrary to the concrete best interests of the child; it must coexist, within the "comparative assessment" of the "concrete" best interest of the child, with other relevant variables, such as – among others – social parenthood, the duration of the relationship that has been established with the child and thus the feeling of identity already acquired by the latter, and the existence of a legislative ban on surrogacy. Therefore, according to the Court, "in all cases in which genetic identity may differ from legal identity, the requirement to strike a balance between the need to establish the truth and the best interests of the child is apparent from the evolution of the law over time".[64]

Lastly, courts may formally grant protection to the right to know one's origins even when they give priority to the "reproductive will" of social parents over the genetic or biological truth of childbirth. In this case, courts recognise the right to know the conditions of birth (which is a broader concept than genetic identity) and a resulting duty to disclose on the part of the intended parents. This happened in Argentina, where the courts, confronted with a legislative lacuna in the field of surrogacy,

[61] M.R. and D.R. (suing by their father and next friend O.R.) &ors -v- An t-Ard-Chláraitheoir&ors, [2014] IESC 60, 7 November 2014.

[62] Denham C.J., § 113-119; see A. MARGARIA, *Nuove forme di filiazione e genitorialità. Leggi e giudici di fronte alle nuove realtà*, Bologna, 2018, 240.

[63] F. ANGELINI, *Bilanciare insieme verità di parto e interesse del minore. La Corte costituzionale in materia di maternità surrogata mostra al giudice come non buttare il bambino con l'acqua sporca*, in *Costituzionalismo.it*, 1, 2018,149-177.

[64] Italian Constitutional Court, Judgment no. 272 of 2017, cit.

ISSN 2284-4503

have adopted a mechanism of *ex ante* judicial authorisation of surrogacy agreements,[65] which include the duty to inform the child about the circumstances of birth, in the light of respect for the child's right to personal identity.[66] Concretely, the courts systematically ordered that, in safeguarding the right to identity, which is also constitutionally protected, in the event of birth, intended parents must make their child aware of his/her gestational truth, for when he/she is old and mature enough to understand his/her life history.[67] A common line of reasoning is the assertion that children born under a surrogacy agreement, as part of their identity, have the right to know that they were born through the use of surrogacy and have also the right to know the identity of the surrogate mother.[68]

4.3. Surrogacy and the relevance of genetic and biological ties: a multifaceted issue

Within the framework of surrogacy, the role played by the right to know one's genetic origins is decisively oriented by the triplication of the possible forms of motherhood – genetic, biological and social – on the one hand; but it is also directly interested by a distinction within the concept of personal origins, which derives from the combination between gamete donation and surrogacy. Therefore, if the dichotomy between genetic and biological mother usually follows the rules on the anonymity of donor identity or sensitive data set forth in any legal system in the context of assisted reproduction technologies, the role played by the right to know one's genetic or biological origins becomes more complex and unpredictable when the separation between gestational and social parenthood comes into play. Here, legislature is the pivotal authority called to find a reasonable balancing between competing rights and interests, both in cases where surrogacy is regulated and when the law provides for a total ban of such practice. The right to know can be limited similarly to the case of gamete donation, even though we have seen that courts may reverse the anonymity rule in order to guarantee the personal identity of the child born via surrogacy in a more effective way (Portugal). Alternatively, it can be particularly valorised as a form of expression of the best interest of the child, when the law provides that a genetic link with the intended parents must exist for the surrogacy agreement to be lawful (South Africa). If we consider the courts' attitude, it is possible to detect different approaches, which range from an absolute self-restraint with regard to the legislature's margin of appreciation, even if the need for a clear normative framework is explicitly declared (Ireland); to the recognition of the paramount nature of the best interest of the child, within which the need to protect one's genetic identity shall not act as a limit for the effective protection of the broader personal identity of the child born via surrogacy (Italy); and to the definition of a formal duty for the intended

[65] D.M. CASTANO VARGAS, *La procreazione medicalmente assistita. Prospettiva di bilanciamento dei diritti nell'esperienza argentina*, in *Biolaw Journal-Rivista di BioDiritto*, 3, 2018, 215-217.

[66] According to L. POLI, *Artificial reproductive technologies and the right to the truth about genetic and biographic origins*, cit., 60, "Mutatis mutandis, a similar principle is applicable in the case of surrogacy, especially considering that prenatal attachment to the gestational mother might be relevant for the definition of the individual identity".

[67] *R., L. S. y Otros s/ Solicita Homologación* sentencia 22 de Noviembre de 2017 Juzgado de Familia 2da Nominación, Cordoba (SAIJ: FA17160037).

[68] Tribunal Colegiado de Familia Nº 7 de Rosario 5 de diciembre de 2017 *H., M.E. y Otros S/Venias y Dispensas*, 32.

parents to disclose the conditions of birth to the child born via surrogacy, even in the context of a regulatory regime based on the *ex-ante* judicial authorisation of surrogacy agreements (Argentina).

5. Concluding remarks: the right to know one's genetic and biological origins; a relational and multidimensional right

The right to know one's genetic origin has a relational nature, as it must find a balance with other competing individual rights, which belong to donors, to the gestational mother and even to the child him/herself (i.e. the right to a safe birth, as in the case of anonymous birth). It may also be formed by different dimensions, strictly dependent upon the concrete context at stake (anonymous birth, gamete donation, surrogacy) and mainly based on the distinction between genetic and biological identity, which must both be understood as functional and not in competition with the child's right to personal identity and his/her best interest.

With regard to its concrete normative content, the analysis of the framework concerning this right in the context of adoption, anonymous birth, assisted reproduction and surrogacy confirmed that "genetic origin" can be legitimately defined as an "umbrella term", which covers different kinds of information related to donors or the gestational mother. Accordingly, the right at stake may find at least three different forms of legal recognition. The first one is the right to have access to health or genetic information linked to the donors or the biological mother. This is usually enforceable, as it represents the essential core of the right. Secondly, the duty belonging to social parents to disclose the conditions in which birth occurred. As we have seen, this might be hard to enforce, due to the very intimate nature of the relationship between parents and children and family ties. Finally, the highest level of recognition, which corresponds to the right to know the full identity of donors and the gestational mother and which can be considered an exception at the legislative level. As we have seen, it has raised several complex issues in the delicate field of anonymous birth, which gives origin to a special form of adoption.

The relational and multidimensional nature of the right to know one's genetic origins also guides the concrete legal framework designed by the legislature, to which a broad margin of appreciation is usually granted, in order to set a reasonable balance between the competing rights and interests at stake. At the same time, courts may be directly or indirectly involved in the concrete enforcement of the legislative framework. It may happen directly, when the law provides for requirements, criteria or procedures upon which the effective implementation of the child's right related to one's origins is conditioned; or when, from a constitutional perspective, a judicial assessment based on a case-by-case approach is required in order to define the concrete best interest of the child. It may occur also indirectly, when courts are called to assess the legitimacy of legislative choices able to limit or affect the child's right also from the perspective of the protection of his or her biological or genetic identity. In any case, the recognition of the prerogatives linked with the process or characteristics of birth must be functional to the determination of the concrete best interest of the child intended in a dynamic and comprehensive way, in which the right to know may be designed to represent an essential object of protection without becoming an obstacle to the building and the development of the child's personal identity as a whole.

Is it possible to place limits on the self-determination of your own genetic data? Certainly, and there is an urgent need for it!

*Iñigo De Miguel Beriain, Daniel Jove**

ABSTRACT: Voluntary disclosure of data is becoming an increasingly common practice. The problem is that these actions can seriously harm the relatives of those make such disclosures. This could happen with genetic data, which belongs to all persons about whom it provides information, regardless of who the sample donor is. What can be done in this situation? We defend the idea that they are the rights conferred by the GDPR to data subjects. On this basis, any processing of genetic data should be seen as an exercise of balancing interests, except where the need to respect professional secrecy requires otherwise.

KEYWORDS: DTC tests; voluntary disclosure of data; informational self-determination; collective data; data of relatives

SUMMARY: 1. Introduction: Family dinners, direct-to-consumer tests (DTC) and data protection – 2. Data protection, personal data and self-determination rights – 3. Genetic data are the personal data of different subjects – 4. Data protection as a tool for embedding conflicting interests – 5. First objection: The GDPR states that genetic data are only personal data of the sample donor – 6. Second objection: Data could become everyone's data because we are all related genetically – 7. Third objection: Until it is checked, we do not know if it is other people's personal data – 8. Fourth objection: If we accept the hypothesis, the research system would suffer terrible consequences – 9. Conclusion.

1. Introduction: Family dinners, direct-to-consumer tests (DTC)

Christmas Eve dinners are, in most western countries, a good time for family gatherings. They are meetings that often yield wonderful discussions in which, out of affection, the cousin we hardly ever see devotes himself to openly ragging two of his siblings or even his spouse even before dessert is served. There are, however, some years in which a confluence of stars brings about peace and harmony. For those for whom this situation will never be an acceptable scenario, it is more than advisable to bring up a hitherto underused resource to get the wheels turning: express your willingness to publicly disclose your own genetic data by publishing the results of a DTC on a public platform (Facebook, for example). This will display information on the presence of dominant pathological genes in your DNA, and the propensity for certain pathologies, and so on. As we all

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ISSN 2284-4503

share 12.5% of our DNA with our cousins, more so with our parents, descendants or siblings,[1] and these data could be used for very damaging purposes – whether it is to solve a crime, get a job or obtain medical insurance – it is quite likely that our goal of livening up the evening for the grandmother who has been disappointed by an untimely oasis of peace will be adequately fulfilled.

The example we have just given can (and does) happen in a world where DTCs are becoming increasingly common, clearly being a growing business.[2] If we add to this the fact that sharing even the most intimate parts of their lives on social networks has become a way of life for many people, our ability to access sensitive information increases substantially. Just think, some crimes are already being solved thanks to the use of DNA from family members, which has generated some ethical controversy.[3] The day when companies use tracking tools to value the genetic profiles made public by reckless, if not malicious, family members (or others) does not seem far off. Regardless of one's own prudence and rectitude, the indiscretion of others can be just as damaging.

In light of this scenario, there is an urgent need to analyse what we can do to protect the data we share with others, considering the limitations offered by current regulations, at least at European Union (EU) level. This will not be an easy task, as much of the legal discourse has been built based on the empowerment of the individual as an isolated subject. Therefore, it is often easy to arrive at excessive interpretations of the right to self-determination over one's own data. This approach is not the most appropriate to the principles of justice that require consideration of the interests of others in the exercise of one's rights. Furthermore, it is not an inevitable consequence of the application of the existing legal framework. On the contrary, it is possible and appropriate to draw the boundaries of determination for one's own data in accordance with the provisions of the General Data Protection Regulation (GDPR). However, we need to take this scenario seriously and explore the best means of dealing with voluntary disclosure that causes harm to third parties from a legal point of view. This article will be devoted to developing this argument.

2. Data protection, personal data and self-determination rights

Determining how we can defend ourselves against possible attacks on our privacy by third parties with whom we have the (dis)grace of sharing genes is not a simple issue. Voluntary disclosure scenarios place us in the eye of the hurricane of a struggle that confronts two different paradigms. On the one hand, a thought pattern is related to the paradigm of medical consent, which sometimes links the object to be protected – the information – with the subject that provides it, i.e. the sample donor. Based on this belief, it is considered that the right to informational self-determination should practically have no limits, as the data belong to the person who provides it and to no one else. For

[1] *Privacy implications of genetic data sharing*, available at: https://www.ecseq.com/blog/2019/privacy-implications-of-genetic-information-sharing (last visited 07/09/2020).

[2] S. THIEBES, P.A. TOUSSAINT, J. JU, J. AHN, K. LYYTINEN, A. SUNYAEV, *Valuable Genomes: A Taxonomy and Archetypes of Business Models in Direct-to-Consumer Genetic Testing*, in *Journal of Medical Internet Research*, 22, 1, 2020,1-16, DOI: 10.2196/14890.

[3] C.J. GUERRINI, J.O. ROBINSON, D. PETERSEN, A.L. MCGUIRE, *Should police have access to genetic genealogy databases? Capturing the Golden State Killer and other criminals using a controversial new forensic technique*, in *PLoS Biology*, 16, 10, 2018, DOI:10.1371/journal.pbio.2006906.

this reason, in the biomedical field, data are often treated as if it were an exclusive right of the person providing the sample, even though data protection regulations make it difficult to support this interpretation. On this basis, it would be very difficult (perhaps impossible) for relatives to raise an objection to the public display of a person's genetic data, as it is obvious that if the donor decides to publish genetic data from one of their samples, they would be exercising the right of self-determination over their data the law confers on them. This means of understanding the relationship of individuals with their genetic information therefore leads to a dead end.[4]

There is, however, a reasonable alternative to this status quo. However, understanding it means leaving the traditional medical law framework to enter the turbulent waters of data protection law. From this perspective, data are the object of a right that belongs to all subjects affected by the information transmitted, regardless of which, or who, the source is. In the context of the EU, this means taking as an unavoidable reference the GDPR, which regulates everything relating to personal data protection.[5]

Indeed, the paradigm constructed by the GDPR is based on a right: the right to informational self-determination, enshrined in Article 8 of the Charter of Fundamental Rights of the European Union (CFR). By virtue of this, it is the data subjects who decide on the destination of their data. However, this right does not confer unlimited powers on its holder, "but must be considered in relation to its function in society".[6] Actions such as voluntary disclosure must therefore be weighed against the rights and freedoms of other data subjects involved. In order to do so, we must balance the different elements involved in this scenario. To this purpose, two premises must be taken into consideration: 1) Some data may be the personal data of more than one person,[7] and 2) Therefore, if different data subjects express different views on a particular processing of these data (such as their publication on a social network), a conflict of interest – which will have to be resolved in each individual case – occurs. Next, we will explore each of these issues in depth.

[4] As Clayton et al. stated: "one of the most significant challenges is that many people take genetic data about themselves, which they often received from DTC companies, and post them online in an identifiable form to find their relatives, to share with other people with similar conditions, or to promote research. These actions necessarily reveal information about their relatives, as has been made clear by the use of GEDMatch to identity criminal suspects. At present, a person has no ability to prevent his or her relatives from revealing their own information. Moreover, there are no limits on who can access these data or for what purpose". In E.W. CLAYTON, B.J. EVANS, J.W. HAZEL, M.A. ROTHSTEIN, *The law of genetic privacy: applications, implications, and limitations*, in *Journal of Law and the Biosciences,* 6, 1, 2019, 1-36, DOI: https://doi.org/10.1093/jlb/lsz007.

[5] The GDPR is the most complete data protection standard. It offers the better system of guarantees, which makes it a reference model for other countries. Furthermore, thanks to its territorial scope, it is able to condition the processing models of those countries that intend to process data on EU citizens: Case C-362/14, *Schrems v. DP Commissioner*, ECLI:EU:C:2015:650; Case C-311/18, *Data Protection Commissioner v. Facebook Ireland Limited y Maximillian Schrems*, ECLI:EU:C:2020:559, relating to Safe Harbour and Privacy Shield.

[6] Joined Cases C-92/09 and C-93/09, *Volker und Markus Schecke GbR v. Land Hessen y Eifert v. Land Hessen y Bundesantalt fur Landwirtschaft un Ernahrung*, ECLI:EU:C:2010:662, para. 48.

[7] Case C-434/16, *Peter Nowak v. Data Protection Commissioner*, ECLI:EU:C:2017:994, para. 45.

3. Genetic data are the personal data of different subjects

In general, it is often believed that personal data obtained from a biological sample belongs to the donor, who has an almost unquestionable right to decide on the information extracted from the sample – "my sample, my data". Conversely, the donor's relatives are often denied any prerogative over that information.[8] In our view, however, this concept is not compatible with the legal framework drawn up by the GDPR, as it is incompatible with the definition of personal data. Other errors are derived from this original error, such as the failure to recognize the rights (access, rectification, restriction of processing) that the data protection framework confers to the subjects whose personal data are being processed. This fact, on the other hand, ultimately leads to the vulnerability of those who are directly affected by the public disclosure of data obtained from the analysis of a sample that was obtained from a donor different to themselves. Thus, change is truly needed for this perspective. However, to do so, it is necessary to cement the linkage between information about a subject and personal data. This requires a deep understanding of the concept of personal data. Article 4 of the GDPR states that "personal data" means "any information relating to an identified or identifiable natural person ("data subject"); an identifiable natural person is one who can be identified, directly or indirectly, in particular by reference to an identifier such as a name, an identification number, location data, an online identifier or to one or more factors specific to the physical, physiological, genetic, mental, economic, cultural or social identity of that natural person".

The concept of personal data, therefore, is broad and covers all types of information.[9] The key to determining whether the information obtained from a sample is a subject's personal information is whether, "by reason of its content, purpose or effect" that information is linked to a particular person[10] and, in that case, from which person. If it is possible to connect this information with a natural person, this information will be their personal data, without excluding other subjects.

The essential question, in short, is to determine whether the information extracted from a biological sample is personal data not only of the donor, but also of other people related to them. In the case of genetic data, and "to the extent that genetic data has a family dimension, it can be argued that it is "shared" information, with family members having a right to information that may have implications for their own health and future life".[11] They should therefore be considered the personal data of all concerned data subjects. The Article 29 Working Party has stated this, at least indirectly, by considering that the data collected from the samples of deceased people are considered their relatives' personal data, as "the information on dead individuals may also refer to living persons. […] Thus, where the information which is data on the dead can be considered to relate at the same time also to the living and be personal data subject to the Directive".[12]

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[8] P. NICOLÁS, *Los derechos sobre los datos utilizados con fines de investigación biomédica ante los nuevos escenarios tecnológicos y científicos*, in *Revista Derecho y Genoma Humano*, extraord. number, 2019, 129-167.

[9] Case C-553/07, *Rijkeboer*, ECLI:EU:C:2009:293, para. 59.

[10] Case C-434/16, *Peter Nowak v. Data Protection Commissioner*, ECLI:EU:C:2017:994, para. 35.

[11] A29WP, *Working Document on Genetic Data*, adopted on 17 March 2004, 8, available at: https://ec.europa.eu/justice/article-29/documentation/opinion-recommendation/files/2004/wp91_en.pdf.

[12] A29WP, Opinion 4/2007 on the concept of personal data, adopted on 20th June. The ICO has also stated explicitly that, "[i]n the case of requests for the medical records of a deceased person, it is possible that this could

However, if this is true for the deceased, it must also be true for the living, as the information is the same.

Therefore, the real issue is determining whether a data is personal and which individual's information it provides. In this regard, the source of the information – the biological sample, in this case – or the subject from which it was originally extracted is irrelevant (although it will be important for assessing conflicting interests). Thus, it must be concluded that the information obtained from the genetic analysis of donor samples is personal data of these subjects, but is also their relatives' data. Recognizing that certain information is personal data implies that the relatives receive the protection that the right to data protection confers, with the exceptions that the GDPR introduces regarding professional secrecy, which will be explained in the following sections. The question of which interest prevails if their interests diverge is different and will have to be resolved. There are sufficient mechanisms to proceed, as the next section shows. However, we cannot deny that a person has a right only because we do not know how to address the concurrent interests.[13]

4. Data protection as a tool for embedding conflicting interests

We have stated that genetic data might be the personal data of different subjects. This obviously means that there will be different wills involved in making decisions about them, and this means conflict. Therefore, we have to analyse whether this is an unsolvable problem, or whether it can be solved by the currently applicable legal framework. We adhere to the latter, as we believe that it is perfectly possible to resolve conflicts based on the GDPR. In this section, we will explain how.

Resolving conflicts of interest involves different variables that give rise to different scenarios. First, the legality of the controversial processing, that is, its legal basis, must be analysed. When this is not consent, the will of the interested party is not the decisive factor that justifies such treatment. In these cases, the interests of the data controller, the purpose of the processing, the public interest or the legal good to be protected are the elements that justify and condition the processing. At the same time, they are the criteria to be assessed in case of a possible conflict of interests between a data subject (A) who wants her data to be processed and a data subject (B), who has a different intention with regard to the same data (which also refers to her).

The resolution of this type of conflict may seem complex, but in practice it is not, precisely because the legal basis of processing and its conditions provide the necessary elements to carry out the balancing of interests. A wide variety of situations can arise. Let us imagine, for example, that a person performs, and pays for, a DTC, but does not want to share the results with her family members.

include genetic information which may also identify surviving relatives and thereby meet the definition of personal data under the Data Protection Act", available at: https://ico.org.uk/media/for-organisations/documents/1202/information-about-the-deceased-foi-eir.pdf.

[13] In addition, if there is a general interest objective that justifies a limitation of the rights of family members against the subject who provides the sample, it may be articulated by law, as long as the essential content of the right to data protection is respected and the measure is proportional. In this way, the legislator could give protection to certain situations (those in which there is a general interest that is properly justified) while the others are intended for the assessment of the conflicting interests. This action should be carried out, in the first place, by the data controller supported by the data protection officer (if any) and, if the dispute persists, by the supervisory authorities and, ultimately, by courts.

However, one relative, perhaps a clever nephew, decides to make use of the right of access and requests from the company that carried out the DTC that part of the information that may concern him. In this case, we would have, on the one hand, the aunt's interest in keeping secret, perhaps reinforced by a commitment to confidentiality, the company's own business model that could be prejudiced, and on the other hand, the nephew's right of access. In such a case, the right of access would probably not prevail, as the aunt's private life would be a difficult obstacle to overcome unless other reasons were provided in addition to the nephew's mere interest in knowing, not to mention what the GDPR stipulates about the duty to respect professional secrecy, which we detail later.

Let us now imagine that we have a case where the processing is necessary for the concluding a contract between the controller and data subject A, or to protect A's vital interests or for the fulfilment of a legal obligation. For data subject B to be able to prevent processing based on such grounds, her legal assets would have to be affected to an extent sufficient to outweigh the legitimate purpose of the processing. A different issue is data subject B exercising her right of access: this claim would have a better chance of success because it is an instrumental right, while allowing the data subject to exercise other rights,[14] such as the right to rectification (in order to rectify you must first know what information is being processed).

If the legal basis for a particular processing operation were instead the legitimate interest, the decision would be simpler. In these cases, data controllers must always carry out a prior analysis of the conflicting interests, as well as weigh the possible risks and effects on the rights and freedoms at stake. It would only be necessary to ensure that, in this assessment, they have considered the possible existence of more than one data subject with respect to the information with which they are dealing.

The solution, in short, depends on each specific processing. In any case, and in the final analysis, it will always be the data protection authorities or the courts who decide which interest prevails according to the circumstances. In other words, it would be necessary to analyse the different interests involved in each processing operation, the level of affectation of the rights, the purpose of the processing and the context of the processing itself.

Let us now imagine a complex case: the processing is based on the consent of one of the data subjects, the sample donor. In this case, the doubts are overwhelming. First of all, what are the obligations of the controllers? Must they obtain the consent of not only the person providing the information, but also of all those to whom it refers? Even if the answer were negative, even if we think that the consent of the donor is the only necessary consent, the controller would have to address the information duties corresponding to Article 14 of the GDPR. The legal answer, in short, depends on each specific processing. In any case, it will always be the data protection authorities or the courts that will decide which interest prevails in view of the circumstances. In other words, it would be necessary to analyse the different interests involved in each processing operation, the level of affectation of the rights, the purpose of the processing and the context of the processing itself. Therefore, in the case of genetic data, it should at least inform the next of kin (third or fourth degree), as this does not seem to be a disproportionate effort. Complex? Yes, no doubt, but the conflict could be resolved.

[14] Case C-434/16, *Peter Nowak v. Data Protection Commissioner*, ECLI:EU:C:2017:994, para. 57.

Furthermore, should the controller facilitate the exercise of the various rights (access, rectification, objection, etc.) to any data subject or only to the sample donor? If so, how should conflict situations be resolved? Once again, there are no general solutions applicable. In cases such as voluntary disclosure of genetic information on social networks and the eventual request of withdrawal by a relative, the resolution seems clear. The chances of the balance tipping in favour of the relative requesting removal are very high. Genetic data are special category data. Their processing, as a general rule, is prohibited (GDPR Article 9(1)), unless any of the circumstances foreseen in Article 9(2) of the GDPR applies. This means that the data subject who does not want to see this information published loses the additional protection afforded by the prohibition on the processing of GDPR Article 9(1). Of course, there will be situations where there may be reasons to justify such interference with the rights of an individual, but these will be the least. In most cases, the interest of the person who wants to make their genetic information no longer public should prevail. The slightest impairment of fundamental rights – both of the right to data protection and of others such as privacy or health – and the risk of discrimination will operate as reference criteria for elucidating conflicts between data subjects.

In any case, what is proven is that this approach from the GDPR legal framework provides criteria that make it possible to impose limits on some of the data disclosure we are analysing (especially the most disproportionate ones), as well as to bring peace to present and future family celebrations. This is despite the doubts regarding the measures that should be required from the processor or the complexity inherent in resolving any conflict of interest. However, we are aware that our proposal is complex and may generate opposition. For this reason, in the following sections, we analyse and respond to some of the possible criticisms thereof.

5. First objection: The GDPR states that genetic data are only personal data of the sample donor

The first objection to everything we have clarified so far claims that a genetic data is only a personal data of the sample donor because that is what the GDPR rules. This argument is based on Article 4(13) of the GDPR, which states that genetic data are data obtained, "in particular, from an analysis of a biological sample from the natural person in question". Moreover, Recital 34 states that "genetic data should be defined as personal data relating to the inherited or acquired genetic characteristics of a natural person which result from the analysis of a biological sample from the natural person in question, in particular chromosomal, deoxyribonucleic acid (DNA) or ribonucleic acid (RNA) analysis, or from the analysis of another element enabling equivalent information to be obtained".

These definitions, in short, fuel the idea that the information contained in the sample is only genetic data with respect to the sample donor. However, there are reasons for rejecting this option.

The first is that both formulas are not similar. The formula in the Recital emphasizes the origin of the data, but the fact that it ends with the phrase "or from the analysis of another element enabling equivalent information to be obtained" is revealing. It shows that the GDPR focuses on the information itself and not on the source or method of obtaining it. The definition in Article 4(13) reinforces this interpretation, as it does not appear to be exhaustive, but rather exemplary. The use of the

expression "in particular" instead of other options such as "only" and "exclusively" is a clear sign that the EU legislator did not wish to restrict the concept of genetic data to that with that particular origin, but rather to emphasise the more typical method of obtaining it.

Moreover, even accepting the argument that the definition of genetic data in the GDPR is restrictive, this would not deny the information its status as personal data, only the status as genetic data. In other words, we would not say that the data extracted from a biological sample are not personal data, as this conclusion would be incompatible with the definition of personal data, but rather that it would not be genetic data. However, such an interpretation would lead to the consideration that the GDPR would be differentiating two types of DNA-related data: genetic personal data, which would only be associated with the person providing the sample, and non-genetic personal data, that is, data that would provide information about a person, but would not be genetic because it did not originate from that person's sample, although the information would undoubtedly be genetic.

In our view, this interpretation is absurd. Let us imagine that genetic information comes from the sample of a person who has died but has a living twin brother. As is commonly known, this means that they share the same DNA. Thus, the information from one is the same as that from the other. Saying that a piece of information is not the personal data of twin B because it has been obtained from a sample of twin A, despite the fact that the information is equivalent, seems – is – totally incongruous.

In addition, if we review Recital 35 of the GDPR, we find that, among the data that can be considered "personal data concerning health" is "information derived from the testing or examination of a body part or bodily substance, including from genetic data and biological samples". In this case, the GDPR advocates a broader consideration of what is health data. Note that it does not specify from whom the information comes, but talks about "a body part" rather than "his/her body part", and "genetic data and biological samples", not "his/her genetic data and biological samples".

In conclusion, this refutation is not strong enough to be taken into account, although it does at least raise a relevant query: the need to eliminate from the definition the phrase "in particular, from an analysis of a biological sample from the natural person in question", as it only generates confusion. If the legislator were to be embarrassed by such an action, he should at least emphasize its exemplary and not restrictive nature. However, the reason for advocating elimination is that there are currently much more appropriate definitions, such as that in Article 1 of Recommendation No. R (97) 5 on the Protection of Medical Data (February 13, 1997) of the Council of Europe.[15]

[15] Article 1 of Recommendation No. R (97) 5 on the protection of Medical Data (13 February, 1997) of the Council of Europe: "[t]he expression 'genetic data' refers to all data, of whatever type, concerning the hereditary characteristics of an individual or concerning the pattern of inheritance of such characteristics within a related group of individuals. It also refers to all data on the carrying of any genetic information (genes) in an individual or genetic line relating to any aspect of health or disease, whether present as identifiable characteristics or not. The genetic line is the line constituted by genetic similarities resulting from procreation and shared by two or more individuals".

6. Second objection: Data could become everyone's data because we are all related genetically

The second refutation of our proposal is based on the fact that we all share much genetic information with other people, beyond even our relatives. This makes it impractical to consider genetic data as the data of various data subjects. It would lead us to a scenario in which the GDPR could not be applied because eventually any genetic information extracted from an individual could be used to inform judgments about all other humans, and in turn all genetic groups.[16] So, it would be impossible for a data controller to take all groups into account.[17] Therefore, adopting this perspective means distorting the very idea of the right to data protection, which has been built on the basis of the defence of the individual, as a projection of their dignity and free development of their personality, that is, as an individual right, and not as a collective right.

This criticism, once again, is wrong. The GDPR has been applied efficiently to solve problems in which the rights of several data subjects concur on the same data, without resorting to the notion of supra-individual rights. In the case of genetic data, a fundamental factor must also be taken into account: the more distant the biological link, the less information is shared. This means that, in reality, the amount of information on which there may be a conflict will be equal to the percentage of DNA that is shared and the relevance of the information it reveals in each processing. In this way, the data referring to a dominant gene will not be the same as that referring to a recessive one. It will also be necessary to consider whether it is a gene that transmits probabilities of developing a pathology or whether it determines that a data subject will develop with total certainty. Similarly, it is also crucial to know for what the data is used. Processing can have very different consequences for different stakeholders. This evidence must be considered in the resolution of each concrete situation. It will be the context of the processing that "determines or influences the way in which that person is treated or evaluated"[18] and thus their chances of achieving, for example, access to or removal of that information, as discussed in previous sections.

A different – but more complex – issue are cases in which the genetic information extracted from an individual affects a whole community, or what the Article 29 Working Party terms the "biological group".[19] These are cases in which the analysis of an individual's DNA can reveal, for example, information about the lack of immunological resources to address a particular pathology in a community of human beings. Such situations are a more direct challenge of the assumptions of the GDPR, which

[16] E. T. JUENGST, *Groups as Gatekeepers to Genomic Research: Conceptually Confusing, Morally Hazardous, and Practically Useless*, in *Kennedy Institute of Ethics Journal*, 8, 2, 1998, 183-200.

[17] D. HALLINAN, P. DE HERT, *Genetic Classes and Genetic Categories: Protecting Genetic Groups through Data Protection Law*, in L. TAYLOR, L. FLORIDI, B. VAN DER SLOOT, (eds.), *Group Privacy: new challenges of data technologies*, Dordrecht, 2017, 231, available at: https://www.stiftung-nv.de/sites/default/files/group-privacy-2017-authors-draft-manuscript.pdf.

[18] A29WP, *Working document on data protection issues related to RFID technology*, adopted on 19 January, 2005, 8.

[19] A29WP, *Working Document on Genetic Data*, adopted on 17 March 2004, 6, Available at: https://ec.europa.eu/justice/article-29/documentation/opinion-recommendation/files/2004/wp91_en.pdf.

was constructed on the basis of the individual's defence.[20] However, this does not mean that such problems are unmanageable. Rather, it is important to address them as soon as possible. In fact, there are already regulatory precedents in this regard. Article 10 of the UNESCO Universal Declaration on the Human Genome and Human Rights, for example, states that: "[n]o research or research applications concerning the human genome […] should prevail over respect for the human rights, fundamental freedoms and human dignity of individuals or, where applicable, of groups of people".[21] The Article 29 Working Party state that developments in the understanding of genetics may mean a "legally relevant social group can be said to have come into existence – namely, the biological group".[22]

In our opinion, it would be sufficient to generate alternative guidelines to establish inclusion and exclusion criteria: if a data processing from an individual was intended to discover group vulnerabilities, it would be necessary to be particularly attentive to the bases of legitimacy of that processing and to the rights conferred on all affected people. In other words, the characteristics of the processing would condition both its performance and the security measures to be adopted. The impact assessments (Article 35) required by the GDPR are an adequate prevention mechanism to establish a firewall for avoiding undesired situations. In any case, it seems obvious that a calm reflection on the social, ethical and legal problems posed by group profiling is needed. A recent book edited by Linnet Taylor**,** Luciano Floridi and Bert van der Sloot[23] offers an excellent panorama on this issue. We would do well by following up on this basis.

7. Third objection: Until it is checked, we do not know if it is other people's personal data

This rebuttal denies the factual starting point: genetic data only correspond to the sample donors because we can only be sure that they yield reliable information about them and no one else. A similar certainty can only be obtained if a similar genetic analysis of a family member were carried out.[24] Therefore, and as there is no evidence that the information refers to the specific family member in a truthful way, we are not dealing with that family member's personal data.

This refutation, however, is based on the erroneous belief that only data that have actually been proven to relate to a person can be their personal data. In essence, this means accepting the idea that, in order to be personal data, the information must be true. However, there are data processing

[20] Indeed, as Hallinan and de Hert stated, "the link between an individual data subject and their personal data was established on the basis whether the data could identify him or her. Such an approach would be irrelevant in relation to genetic groups". D. HALLINAN, P. DE HERT, *Genetic Classes and Genetic Categories: Protecting Genetic Groups through Data Protection Law*, cit., 231. Available at: https://www.stiftung-nv.de/sites/default/files/group-privacy-2017-authors-draft-manuscript.pdf.

[21] UNESCO, *Universal Declaration on the Human Genome and Human Rights*, 1997, §10.

[22] Article 29, *Data Protection Working Party*, 2004.

[23] L. TAYLOR, L. FLORIDI, B. VAN DER SLOOT (eds.), *Group Privacy: new challenges of data technologies*, Dordrecht, 2017, available at: https://www.stiftung-nv.de/sites/default/files/group-privacy-2017-authors-draft-manuscript.pdf.

[24] P. NICOLÁS, *Los derechos sobre los datos utilizados con fines de investigación biomédica ante los nuevos escenarios tecnológicos y científicos*, cit., 138, available at: https://www.bigdatius.com/wp-content/uploads/2019/12/05_Los_derechos_sobre_los_datos.pdf.

operations that produce effects on a person even if they contain erroneous information. As the Article 29 Working Party stated, "for information to be 'personal data', it is not necessary that it be true or proven. In fact, data protection rules already envisage the possibility that information is incorrect and provide for a right of the data subject to access that information and to challenge it through appropriate remedies".[25]

So, for example, if an insurance company can use a father's genetic analysis to make decisions about his children, even though they know it may not be accurate – they may not be genetically his – that information is the children's personal data (in addition to that of the father's) because it effectively determines how they are treated. As the European Court of Justice (ECJ) has noted, content, purposes and effects are factors that can determine the personal data status of a given piece of information, insofar as they connect it with a specific person on whom they project its consequences[26].

Accuracy or truthfulness are therefore not a precondition for the consideration of information as personal data. Whether the data protection regulations provide for remedies to rectify erroneous information (accuracy principle and right to rectification) is a different matter. What is obvious in any case is that in order to modify them, we must first accept that they are personal data. Otherwise, they would remain in a legal limbo that would be extremely detrimental to the data subjects, as such data would generate effects but we would have no mechanisms for correcting them. The essential point, in short, is that this information "is used to determine or influence the way in which that person is treated or evaluated".[27] If this is the case, then we are referring to personal data. And Thus, as can be understood, and as the examples that have been used throughout this paper demonstrate, genetic data can produce effects beyond that on the donor of the sample from which the information originates. Therefore, this refutation is clearly feeble.

8. Fourth objection: If we accept the hypothesis, the research system would suffer terrible consequences

The last refutation we analyse argues that we should dismiss the idea that data from a sample are personal data of the donor's relatives because its practical effects would be untenable: the use of data for health care or for research would become impossible. As there are multiple data subjects, it would be necessary to ask not only for the consent of the sample donor, but also that of all the other subjects who could be affected by the information gathered, which would greatly complicate the research. Similarly, patients could refuse to undergo tests necessary for preserving their health if they believe that such information could be provided to their relatives.[28]

[25] A29WP, Opinion 4/2007 on the concept of personal data, adopted on 20 June, 2007, 6, available at: https://ec.europa.eu/justice/article-29/documentation/opinion-recommendation/files/2007/wp136_en.pdf.

[26] See Nowak case. In doctrine, S. WACHTER, B. MITTELSTADT, *A Right to Reasonable Inferences: Re-Thinking Data Protection Law in the Age of Big Data and AI*, in *Columbia Business Law Review*, 2, 2019.

[27] A29WP, Working document on data protection issues related to RFID technology, adopted on 19 January, 2005, 8.

[28] C. GIL, *Utilización de muestras biológicas de origen humano con fines de investigación*, in *Revista de Bioética y Derecho*, 25, 2012, 19-32, available at: http://www.ub.edu/fildt/revista/pdf/RByD25_ArtGil.pdf.

There are several reasons, however, to consider that this objection is also inconsistent. To begin, it is not true that considering that the data of a sample are the personal data of the sample donor's relatives implies that they share the same rights as the sample donor. Article 14(5)(d) of the GDPR excludes the obligation to transmit information on the processing to data subjects "where the personal data must remain confidential subject to an obligation of professional secrecy regulated by Union or Member State law, including a statutory obligation of secrecy". Therefore, professional secrecy operates as an instrumental guarantee of the right to data protection and allows for the circumvention of the obligation to inform the relatives in cases of medical diagnosis or treatment. This protection also extends to the case of biomedical research, insofar as it would also be protected by the professional secrecy of those who provide the samples.[29]

This is reinforced by the provisions concerning the processing of special category data (including genetic data and health data), at least in the cases that fall under the circumstances of GDPR Article 9(2)(h). That is, treatments "necessary for the purposes of preventive or occupational medicine, for the assessment of the working capacity of the employee, medical diagnosis, the provision of health or social care or treatment or the management of health or social care systems and services on the basis of Union or Member State law or pursuant to contract with a health professional and subject to the conditions and safeguards referred to in paragraph 3".

It is true that the lack of requirement to inform is limited to activities subject to the duty of secrecy, i.e. care or biomedical research. This means that when transfers are made to third parties or processing is carried out for other purposes, all data subjects must be taken into consideration, informed and their consent obtained[30] – if that is the basis for the original processing. But this is not, in our view, a problem, but rather the opposite: an essential mechanism for limiting the lack of access to information exclusively to cases where there is a clear justification for it. In all other cases, it is perfectly reasonable – and legally enforceable – to inform all those affected of the processing.

We must therefore banish the fear that inspires the objection that we are analysing. As stated earlier, the circumstances of the processing matter. They determine how the various interests involved in each processing operation are addressed. Therefore, not every processing of genetic data will auto-

[29] An example of this secret requirement is the article 5.4 of the Spanish Act 14/2007, 3 July, Biomedical Research. Translation: "Any person who, in the exercise of his or her duties in relation to medical care or biomedical research, to whatever extent, has access to personal data shall be bound by the duty of secrecy. This duty shall continue to apply even after the research or activity has ceased". Original text: "Quedará sometida al deber de secreto cualquier persona que, en el ejercicio de sus funciones en relación con una actuación médico-asistencial o con una investigación biomédica, cualquiera que sea el alcance que tengan una y otra, acceda a datos de carácter personal. Este deber persistirá aún una vez haya cesado la investigación o la actuación".

[30] In this sense, the article 5.2 of the Spanish Act 14/2007, 3 July, Biomedical Research, recognizes as interested parties the relatives of the source subject and establishes that their consent must be obtained for the transfer of information to third parties. Translate: "The transfer of personal data to third parties outside of the medical-healthcare activity or biomedical research will require the express written consent of the interested party. In the event that the data obtained from the source subject could reveal personal information about their relatives, the transfer to third parties will require the express written consent of all concerned". Original text: Art. 5.2: «La cesión de datos de carácter personal a terceros ajenos a la actuación médico-asistencial o a una investigación biomédica, requerirá el consentimiento expreso y escrito del interesado. En el supuesto de que los datos obtenidos del sujeto fuente pudieran revelar información de carácter personal de sus familiares, la cesión a terceros requerirá el consentimiento expreso y escrito de todos los interesados".

matically entail an obligation to transmit that information to all potential data subjects. If consent is the legal basis for data processing, considering that those data would also be the relatives' personal data does not mean that the researcher would need to obtain their consent to perform the research. In principle, medical and professional secrecy guarantees that the actual processing can be carried out, based on the exclusive consent of the sample donor, without infringing the rights of any data subjects. As the GDPR states, a major interest – medical secrecy – rules.

This is obviously the general framework provided by the GDPR. However, that there are cases in which the circumstances render it necessary to disregard such a confidentiality and to transfer certain information to an interested party other than the donor, as may be the case with AIDS patients, cannot be ruled out. In such a scenario, there was conflict between professional secrecy and the need to preserve the health of third parties, a conflict that was resolved in favour of the latter: if revealing secrecy meant that patients would refuse to undergo diagnostic tests, it would be a risk to be assumed, but it would be an imposition on the health of third parties. It is true that the case of genetic data is much more controversial,[31] but in our opinion, the conclusions should be similar. In fact, there are rules that provide that if the analysis of a sample reveals relevant information about the health of the sample donor's relatives, there is an obligation to communicate this information, even if the donor objects. It is even worth recalling some famous (and old) judgements, such as the case of 1999, in which the Italian Guarantor for the Protection of Personal Data allowed access to relevant genetic information to the descendant of a deceased person who had explicitly opposed it, considering that her right to health prevailed over the right to privacy of the deceased.[32]

Therefore, in each case, the different rights in conflict and the entity by which they may be affected will determine the answer. However, there is no doubt that, for the purposes of biomedical research, the GDPR offers a starting point that provides a sufficient level of confidence to not put the research system at risk.

9. Conclusion

If we are to draw any conclusions from this paper, the main one should be this: it is perfectly possible to defend ourselves against misuse of the right to self-determination over data that endangers our privacy. Nonetheless, this is the case only if we accept that a person's genetic data are undoubtedly their personal data, but also of all persons about whom they transmit information that might influence the manner in which that person is treated or evaluated.[33]

Based on this evidence, each processing operation – including that allowing a sample donor to disclose their data publicly – will require specific analysis, as the GDPR requires data controllers to take into account "the nature, scope, context and purposes of the processing operation and the risks of

[31] M. ROTHSTEIN, *Reconsidering the duty to warn genetically at-risk relatives*, in *Genetics in Medicine*, 20, 2018, 285-290, DOI: https://doi.org/10.1038/gim.2017.257; E.W. CLAYTON, B.J. EVANS, J.W. HAZEL, M.A. ROTHSTEIN, *The law of genetic privacy: applications, implications, and limitations*, cit., 1-36.

[32] Garante per la Protezione dei Dati Personali¸ *Dati inerenti allo stato di salute - dati genetici, Cittadini e società dell'informazione*, 1999 (8), 13-15, available at: https://bit.ly/3ezTYnc.

[33] A29WP, *Working document on data protection issues related to RFID technology*, adopted on 19 January, 2005, 8.

varying degrees of probability and gravity to the rights and freedoms of natural persons".[34] The right to data protection is sufficiently flexible to allow the resolution of the different conflicts that may arise, applying precisely the criteria that should inspire the design of all processing. In short, the legal debate should not be on the nature of the information, nor on the right to be applied, but on how to reconcile this evidence with the legal assessment of the legitimacy of the processing that may affect data subjects with opposing interests. However, this requires courageous action.

Some years ago, the Article 29 Working Party stated: "a new, legally relevant social group can be said to have come into existence – namely, the biological group, the group of kindred as opposed, technically speaking, to one's family. Indeed, such a group does not include family members such as one's spouse or foster children, whereas it also consists of entities outside the family circle – whether in law or factually – such as gamete donors or the woman who, at the time of childbirth, did not recognise her child and requested that her particulars should not be disclosed – this right being supported in certain legal systems. The anonymity granted to the latter entities raises a further issue, which is usually dealt with by providing that the personal data required for genetic testing be communicated exclusively to a physician without referring to the identity of the relevant individual. Given the complexity of the issues described above, the Working Party takes the view at this stage that consideration should be given to a case-by-case approach in deciding how to address possible conflicts between the interests of the data subjects and those of their biological family".

After all this time, we still do not have a legal solution for the conflicts that arise from the fact that genetic information provides relevant data about different people. In this scenario, the lack of regulation can lead to clearly abusive behaviour, where the right of self-determination over data is overly protected. In our opinion, this requires urgent intervention, which would not so much require a regulatory reform as a decisive application of the provisions of the GDPR. In particular, it requires the assumption that certain information can be the personal data of more than one person. This should be accompanied by the definitive incorporation of the content, purposes and effects criteria as the basis for identifying certain information as a person's personal data.

With these guidelines as a basis, all those affected by information relevant to their lives would have the status of data subjects and the rights that are inherent to that status. As has been shown throughout this work, the obstacles and objections that could be raised are perfectly surmountable. In short, it is not so much a question of innovating as one of having the courage to apply the rights and regulatory provisions we already have. Let us hope that we have the confidence and courage to use them.

[34] GDPR, Article 24(1).

ISSN 2284-4503

Special issue

You can't put the genie back in the bottle: On the legal and conceptual understanding of genetic privacy in the era of personal data protection in Europe

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ABSTRACT: This article sheds a light on how the data protection requirements enshrined in the General Data Protection Regulation (GDPR) relate to shaping genetic privacy in the context of a complex and integrated genetic testing enterprise. It suggests that the informational dimension of genetic privacy in the era of data protection could be described as a sphere of controlled access. Given that the GDPR does not prescribe quantitative or contextual limitations relating to access once the applicable requirements are met, one could argue that there are good preconditions for the field to head in the direction of genetic transparency. This puts on the agenda the questions of what challenges this could bring and whether adequate mechanisms exist to deal with them.

KEYWORDS: Genetic data protection; genetic privacy; genetic testing; scientific publication; scientific research

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1. Introduction

Genetic privacy has been a major concern in the governance of genetic and genomic technology on both sides of the millennium.[1] The great strides that have been made in science and technology in the last decades of the past century created new possibilities and opportunities for society and different groups of individuals within it, and enabled enhanced access to genetic data and information. This, coupled with the particular nature of genetic information and diverse potential interests in this information, has brought about questions, concerns and risks for individuals and societal groups. They include questions regarding reconciliation of the various interests at stake, and degrees of tolerable interventions, concerns over confidentiality and adequate protection of personal data, risks of discrimination and stigmatisation, and misuse of information in such contexts as employment and insurance.[2] Issues such as these are relevant not only in regard to genetic testing stricto sensu but also in the number of steps that can often be lawfully taken in addition to the testing initially done in a clinical or non-clinical setting. These steps include but are not limited to different activities in the context of furthering scientific developments, such as scientific research and publication.

From the outset, it seems that genetic privacy has increasingly been protected despite the legal and scholarly controversy in the field and disagreement regarding whether and to what extent genetic information differs from other types of information, and therefore merits special protection.[3] In Europe, even early on genetic privacy was subjected to the health and biomedicine legal frameworks, such as to those regulations addressing medical or biomedical interventions generally or genetic testing specifically, or biobanking and data banking or biomedical research. It has also been safeguarded through the protection of the right to private life. More recently, it has been expressly addressed through stringent data protection regimes within both of the European legal orders, the Council of Europe and the European Union.[4] One of the most recent and most comprehensive data protection tools is the EU General Data Protection Regulation (GDPR),[5] a directly applicable legal instrument with far-reaching material and territorial scope of application that has had a profound and extensively discussed, albeit fully unchartered, impact on how genetic privacy is safeguarded in the EU.[6] Alt-

[1] For example, as of 12 February 2021, Pubmed alone has 2,998 results dating back to 1973. Keyword: "genetic privacy". www.pubmed.ncbi.nlm.nih.gov/?term=genetic+privacy&timeline=expanded (last visited 12/02/2021)

[2] Such as employment and insurance. *See*, for example, L. GOSTIN, *Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers*, in *American Journal of Law & Medicine,* 17, 1991, 109.

[3] For example, T. H. MURRAY, *Genetic Exceptionalism and "Future Diaries": Is Genetic Information Different from Other Medical Information*, in M. A. ROTHSTEIN (ed.), *Genetic secrets: Protecting Privacy and Confidentiality in the Genetic Era*, New Haven, 1997, 60–73. *See* further section 2.

[4] *See* Section 2.4.

[5] Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation) OJ L 119, 4.5.2016, 1–88.

[6] Considerable scholarly focus has been placed on biobanking and data banking, *see*, for example, S. SLOKENBERGA, O. TZORTZATOU, J. REICHEL (eds.), *GDPR and Biobanking. Individual Rights, Public Interest and Research Regulation across Europe*, Cham, 2021, and D. HALLINAN, *Protecting Genetic Privacy in Biobanking through Data Protec-*

hough the Council of Europe has followed this trend and has also strengthened its data protection system and the status of genetic information,[7] the central instrument – revised Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data – has not entered into force yet.[8]

One cannot fail to note that over the course of time the Council of Europe and the EU have created several parallel, albeit internally and externally interacting and overlapping mechanisms, that are relevant to protecting genetic privacy. Nor that the GDPR has shaken the scientific research field and sparked debates over scientific research regulation and the protections data subjects have.[9] Given these legal advances and in particular the prominent role that genetic data have received in data protection regimes, one might question how it relates to safeguarding genetic privacy.

This article places the above-mentioned question at its core and aims to shed light on genetic privacy in the era of data protection. Through a theoretical framework of genetic privacy and a selection of distinct steps in the genetic analysis enterprise, this article explores how the data protection requirements enshrined in the GDPR relate to genetic privacy protection measures set out in the health and biomedicine regulatory instruments. As the ambition is to provide an insight into genetic privacy in the era of data protection, two important limitations need to be mentioned. First, the selection of legal instruments and issues is limited to those found in the two European legal orders and is illustrative rather than exhaustive. Second, examination of genetic privacy in the context of data protection under the different steps of genetic testing as a complex enterprise requires accounting for different technologies and techniques, and their applications. They raise diverse and profound ethical, social and legal issues in themselves and have been subject to considerable and ongoing discussions. However, because of the focus of this article and practical limitations I am not able to account for these discussions here in a manner that will do justice to the respective questions and attributable complexities, and neither is this the place to attempt to contribute to the discussions on substantive issues raised by different techniques and technologies, and their applications.

tion Law, Oxford, 2021. Currently, there is a considerable gap regarding empirical studies on challenges and solutions the GDPR has brought.

[7] *See*, for example, Protocol amending the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data, ETS 223 (consolidated), and Committee of Ministers, Recommendation CM/Rec(2019)2 Protection of health-related data.

[8] Details of Treaty No. 223, www.coe.int/en/web/conventions/full-list/-/conventions/treaty/223 (last visited 13/04/2021).

[9] *See*, for example, C. STAUNTON, S. SLOKENBERGA, D. MASCALZONI, *The GDPR and the Research Exemption: Considerations on the Necessary Safeguards for Research Biobanks*, in *European Journal of Human Genetics*, 27, 2019, and D. PELOQUIN, M. DIMAIO, B. BIERER, M. BARNES, *Disruptive and avoidable: GDPR challenges to secondary research uses of data*, in *European Journal of Human Genetics,* 28, 2020.

2. Establishing the foundations: scientific advances, the complexity of a genetic testing enterprise and conceptual and legal foundations of genetic privacy

2.1. Scientific advances

Genetic privacy is a concept that is difficult to analyse without also considering the depth and breadth of possible genetic interventions and the pace of technological and industrial developments in comparison with the legal protections. Instead of attempting an exhaustive account of the scientific, technological and industrial milestones, this section briefly pins down some essentials that show the pace of development, and the depth and breadth of possible genetic privacy interventions. In doing so, it sets the context and shape of the analysis to come.

The completion of the Human Genome Project (HGP) was a turning point in the history of human genetics and genomics. However, several significant steps had paved the path for the project.[10] These include identification of a nuclein in 1869,[11] rediscovery of Mendel's laws in the spring of 1900, and Watson and Crick's elucidation of the DNA molecule in the form of a three-dimensional double helix. They also include a series of milestones in the 1970s, including the emergence of recombinant DNA technology that enabled the joining of DNA of different species,[12] thus opening up opportunities for analysing and modifying gene structure and the organisation of complex genomes.[13] The development of efficient DNA cloning methods enabled scientists to embark on the study of the structure of selected fragments of DNA (e.g. of single genes).[14] At that time, the focus of the study was on a few thousand base pairs and recording longer and longer stretches of DNA in an attempt to deliver a better understanding of the biomolecular function. The discovery of DNA sequencing by Sanger and Gilbert independently enabled scientists to read the genetic code.[15] The early 1980s brought with it the discovery of polymerase chain reaction (PCR), a technology enabling amplification of DNA, thus facilitating its study.[16] Around this time positional cloning also emerged, a technique that became the standard way of finding disease-related genes.[17] During this time, the application of the existing advances focused on, for example, evaluating Mendelian disorders, birth defects and chromosomal

[10] D. J. GALAS, S. J. MCCORMACK, *An Historical Perspective on Genomic Technologies*, in *Current issues in molecular biology*, 5, 2003, 123. *See* also F. S. COLLINS, *Implications of the Human Genome Project for Medical Science*, in *JAMA* 2001, 285, 540. R. A. GIBBS, *The Human Genome Project Changed Everything*, in *Nature Reviews Genetics,* 21, 2020, 575.

[11] L. PRAY, *Discovery of DNA Structure and Function: Watson and Crick*, in *Nature Education*, 1/1, 2008, 100.

[12] National Human Genome Research Institute, *First Recombinant DNA*. https://www.genome.gov/25520302/online-education-kit-1972-first-recombinant-dna (last visited 08/02/2021). S. WRIGHT, *Recombinant DNA Technology and Its Social Transformation 1972-1982*, in *Osiris*, 2, 1986, 2303. *See* also H. HOWARD, E. NIEMIEC, A. SOULIER, *D2.1: State of the art review of human genomic technologies*, SIENNA (2018). https://doi.org/10.5281/zenodo.4067912 (last visited 16/02/2021).

[13] P. BERG, J. E MERTZ, *Personal Reflections on the Origins and Emergence of Recombinant DNA Technology*, in *Genetics*,184, 2010, 9.

[14] D. J. GALAS, S. J. MCCORMACK, *op. cit*., 124.

[15] J. M. HEATHER, B. CHAIN, *The Sequence of Sequencers: The History of Sequencing DNA*, in *Genomics*, 107, 2016, 1.

[16] D. J. GALAS, S. J. MCCORMACK, *op. cit*., 124.

[17] D. J. GALAS, S. J. MCCORMACK, *op. cit*., 124. M. BOBROW, A.H. GRIMBALDESTON, *Medical Genetics, the Human Genome Project and Public Health*, in *Journal of Epidemiology & Community Health*, 54, 2000, 645.

anomalies. From a genetic privacy perspective, although considerable scientific and technical steps were made to improve access to the genome and enhance knowledge about inheritance, privacy interventions remained rather focused and thus smaller in scale.

The HGP began in 1990 and hit a milestone in 2001 with the reporting of the first draft of the human genome. This was completed in 2003, which provided free and universal access to the sequence of the entire genome. In parallel with this, there was a revolution in computation. These advances were rapidly incorporated into the framework of biology.[18] The HGP also brought a new approach to studying DNA, namely, large-scale studies of genome sequences using high-throughput technologies and collecting large quantities of genetic data.[19] Since the HGP, sequencing technologies have advanced considerably,[20] allowing everyone enhanced access to their genome. Alongside these early developments, there was also an increase in understanding about the human genome. Taken together, the early historical milestones and the advances that have occurred in the last few decades have led to the availability of powerful technology to examine the human genetic makeup and deliver an increased understanding of an individual's genetic inheritance, existing and possible risks, diseases and traits. This intervention is currently limited to the existing and continuously evolving understanding about the human genome, the role of different genes, and the interplay between genes and other factors, such as the environment.

The scientific and technical advances have not only been applied in scientific research but also in clinical and personal care.[21] The exact services that are available differ between health systems and service providers. For example, it is now possible to carry out genomic analysis, including sequencing the whole genome, at an affordable cost within a clinic and offer this service directly to consumers. This technology can be applied in non-reproductive as well as reproductive medicine. Moreover, non-medical applications have also emerged.[22] Advances are expected to continue as considerable investments have been made to develop the field further and improve medical care, for example, through personalised medicine.[23] From the genetic privacy perspective, this means increasingly easy, often also increasingly affordable, deep privacy penetration.

[18] R.A. GIBBS, *op. cit.*

[19] F. S. COLLINS, E. D. GREEN, A. E. GUTTMACHER, M. S. GUYER on behalf of the US National Human Genome Research Institute, *A Vision for the Future of Genomics Research,* in *Nature* 422, 2003, 835. E. D. GREEN, M. S. GUYER, *Charting a Course for Genomic Medicine from Base Pairs to Bedside,* in *Nature*, 470, 2011, 204.

[20] P. TYAGI, M. BHIDE, *History of DNA Sequencing*, in *Folia Veterinaria*, 64, 2020, 66.

[21] For example, already by 2005 a study had identified 13 websites offering health-related genetic testing for direct purchase by the consumer. CHERYL BERG, KELLY FRYER-EDWARDS, *The Ethical Challenges of Direct-to-Consumer Genetic Testing*, in *Journal of Business Ethics*, 77, 2008, 17.

[22] E.g. testing for traits 23andMe, "See Our List of Personalised Genetic Reports - 23andMe Europe". www.23andme.com/en-eu/dna-reports-list/ (last visited 12/02/2021). About the use of 23andMe and other commercial genealogy databases for the so-called "forensic genetic genealogy", and the emerging ethical and legal concerns, see G. FORMICI, *From "familial searching" to "forensic genetic genealogy": new frontiers – and challenges – of DNA analysis in criminal investigations*, in this *Special Issue*.

[23] For example, the EU alone allocated €3.276 billion for the years 2014-2020 to advance innovative medicines. "The IMI Funding Model" (IMI Innovative Medicines Initiative). www.imi.europa.eu/about-imi/imi-funding-model (last visited 8/02/2021).

2.2. Complexity of a genetic testing enterprise and diversity of interests

The field of human genomics is not only a highly sophisticated and complex technical enterprise but also a platform where various interests meet and different tensions between genetic privacy and other rights and interests emerge. What interests emerge in a particular case often depend on different factors, for example, the purpose of testing and the particular context in which it is carried out, and what steps are or could subsequently be taken in the context of the care or service provided. Likewise, they depend on other factors, e.g. the subject of concern and the subject's perspective – whether that of the patient, consumer, care-giver, commercial actor or public health system.

To illustrate, the person being tested generally has an interest in obtaining genetic information in relation to the purpose of the test. For example, information obtained as a result of genetic testing can lead to better predictive medicine, improved diagnostics, and better medical and personal, including reproductive, decision-making. However, it can also bring risks, such as contributing to confusion, anxiety and even misguided decision-making.[24] More recently, questions have emerged over incidental findings and identification of variants of uncertain significance and therefore the potential to obtain information other than that being targeted in the course of primary genetic testing,[25] and how these should be managed to reconcile different interests at stake and safeguard the rights and interests of the person being tested, including an eventual wish not to know.[26] Because of the familial nature of genetic information, there could also be a potential interest for family members to be informed about their individual risks that have become known as a result of a relative's testing.[27] The familial nature of genomic information therefore triggers questions such as how to balance the privacy of the person being tested against the interests of, and even potential duties toward, the relatives of that person.[28] Finally, and by way of illustration, one can note that information obtained as a result of genetic testing could also be of interest to others, for example, employers and insurers,[29] and thus one could question whether and under what circumstances such interventions in genetic privacy, given the risks to the individual, could be acceptable.

The clinician and geneticist generally have an interest, often expressed as a legal duty, in providing qualitative care to a patient.[30] The testing service provider is commonly interested in pursuing its business interests associated with the provision of the service conceptualised under the property

[24] Points such as these have particularly been raised in connection to direct-to-consumer genetic testing.

[25] B. HOFMANN, *Incidental Findings of Uncertain Significance: To Know or Not to Know - That Is Not the Question*, in *BMC Medical Ethics*, 17, 2016, 13.

[26] On the topic see A.O. COZZI, *Incidental Findings and the Right Not to Know in Clinical Settings: Constitutional Perspectives*, in this *Special Issue*.

[27] B. M. KNOPPERS, *Genetic Information and the Family: Are We Our Brother's Keeper?*, in *Trends in Biotechnology*, 20, 2002, 85.

[28] *See*, for example, S.M. SUTER, *Whose Genes Are These Anyway?: Familial Conflicts over Access to Genetic Information*, in *Michigan Law Review*, 91/1854, 1993.

[29] *See*, for example, L. GOSTIN, *op. cit. See* also B. A. LENOX*, Genetic Discrimination in Insurance and Employment: Spoiled Fruits of the Human Genome Project*, in *University of Dayton Law Review,* 23, 1997, 189.

[30] UN Committee on Economic, Social and Cultural Rights (CESCR), *General Comment No. 14: The Right to the Highest Attainable Standard of Health (Art. 12 of the Covenant)*, 11 August 2000, E/C.12/2000/4, para.12.

rights.[31] Both the provider and care-giver could have additional interests in scientific research and the advances it could lead to for such purposes as quality improvement and enhanced care availability and accessibility, and for pursuing relevant collaborations in that regard, as well as disseminating scientific research results. Scientific advances and new applications are also in the interest of society generally.[32] In some situations, the interests are overlapping or even colliding. It is precisely the areas of collision that raise questions over reconciliation of these interests at stake and the degree of tolerable intervention in genetic privacy for the purposes of safeguarding other rights or interests.

2.3. Conceptual foundations of genetic privacy

Although the notion of privacy has a considerable contemporary legal history,[33] genetic privacy is relatively new. It emerged in the late 20th century in response to the scientific and technological advances in the area of genomics and the risks that these advances could bring for individuals. As Laurie put it, "[t]here was no such concept as genetic privacy before scientific advances provided us with the means to gather and manipulate genetic information."[34] However, about three decades later, at the beginning of 2021, and in light of the significant legal advances in the field, the topic has undergone some transformation and now the focus appears to be on the challenges of enhanced data protection requirements, and in particular the challenges that the GDPR has brought to Europe and beyond.

At the core of the early discussions on genetic privacy was often questions about how qualitatively different genetic information is from other types of health information and whether it should be protected in any special way, and if so, how.[35] It was usually conceptualised under the umbrella term of genetic exceptionalism, which gave rise to a rather polarised debate. The opponents often argued that genetic information in many ways is comparable with other (usually health) information, and could be just as sensitive as other information.[36] The proponents often acknowledged overlaps between genetic information and other types of information, and that different genetic information could have different degrees of sensitivity.[37] However, the uniqueness attributable to it, including the concentration of a multitude of personally identifiable facts in a single (and portable) biological material,[38] as well as the depth of possible intervention, supported enhanced protection.[39]

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[31] e.g. Convention for the Protection of Human Rights and Fundamental Freedoms, ETS 005, Protocol 1, Article 1.

[32] As protected under, for example, Article 15 of the International Covenant on Economic, Social and Cultural Rights and Article 27 of the *Universal Declaration of Human Rights*.

[33] On the emergence of privacy as a human right, *see* O. DIGGELMANN, M. N. CLEIS, *How the Right to Privacy Became a Human Right*, in *Human Rights Law Review*, 14, 2004, 441.

[34] G. LAURIE, *Genetic Privacy: A Challenge to Medico-Legal Norms*, Cambridge, 2002, 25.

[35] *See* T. H. MURRAY, *op. cit*. For a review of these differences, *see* section 2 in L. O. GOSTIN, J. G. HODGE, *Genetic Privacy and the Law: An End to Genetics Exceptionalism*, in *Jurimetrics*, 40, 1999, 21.

[36] *See* e.g. T. SPAAK, *Genetic Discrimination*, in *Minnesota Journal of Law, Science and Technology*, 7, 2005, 639. Some have also argued that there exists several interests that lie behind the movement towards exceptionalism, notably money and fame. W. BAINS, *Genetic Exceptionalism*, in *Nature Biotechnology*, 28, 2010, 212.

[37] M. RICHARDS, *How Distinctive is Genetic Information?,* in *Studies in History and Philosophy of Science Part C: Studies in History and Philosophy of Biological and Biomedical Sciences*, 32/4, 2001, 663.

[38] L. GOSTIN, *op. cit.*, 36.

Conceptually, genetic privacy has often been understood as a privacy right, though considerations of other rights, e.g. property rights, have also been put forward as a means to protect privacy.[40] If genetic privacy is located under the protection of privacy then, at least theoretically, genetic privacy could have as many diverse understandings as privacy itself.[41] To illustrate, privacy has been approached as the right to be left alone,[42] as limited access to the self,[43] as secrecy,[44] as control over personal information,[45] as personhood,[46] and as intimacy.[47] Several more concrete approaches to genetic privacy have also been put forward.[48] For example, some have focused on its informational aspect and the control in that regard;[49] others have argued that it is a multidimensional concept consisting of the following facets – informational privacy, physical privacy, decisional privacy and proprietary privacy.[50] In the context of genetic testing, Laurie has defined genetic privacy as a state of separateness.[51] Such a definition serves to distinguish the private from the public and demarcates zone that shields individuals from unwarranted inclusions in zones of protection. For Laurie, in the context of genetic analysis, this state of separateness consists of two elements, spatial and informational privacy. Spatial privacy focuses on the "state of non-access to the individual's physical or psychological self", whereas informational privacy focuses on the "state in which personal information about an individual is in a state of non-access from others".[52] From this perspective, information that is returned to the person being tested seems to relate to spatial privacy,[53] whereas information shared with others to informational privacy.

One of the central pillars in the context of genetic information from the perspective of an individual has been autonomy, expressed as the "right" to control one's genetic information.[54] How exactly autonomy relates to genetic privacy, whether those are two distinct approaches or whether autonomy

[39] *See*, for example, ECtHR reasoning in the case of *S and Marper v United Kingdom* (Application No. 30562/04, 4 December 2008).

[40] For example, R. A. SPINELLO, *Property Rights in Genetic Information*, in *Ethics and information technology*, 6, 2004, 29. C. M. V. BARRAD, *Genetic Information and Property Theory*, in *Northwestern University Law Review*, 87, 1992, 1037. S. M. SUTER, *Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy*, in *George Washington Law Review*, 72, 2003, 737.

[41] For a thorough review of different approaches to privacy *see* D. J. SOLOVE, *A Taxonomy of Privacy*, in *University of Pennsylvania Law Review*, 154, 2005, 477.

[42] E.g. S.D. WARREN, L. D. BRANDEIS, *Right to Privacy*, *Harvard Law Review*, 4/193, 1890-1891.

[43] E.g. R. GAVISON, *Privacy and the Limits of Law*, in *The Yale Law Journal*, 89, 1980, 428.

[44] E.g. R. A. POSNER, *Economic Analysis of Law*, Boston, 1973, 43.

[45] E.g. A.F. WESTIN, *Privacy and Freedom*, New York, 1967, 7.

[46] E.g. J. BRAXTON CRAVEN, *Personhood: The Right to Be Let Alone*, in *Duke Law Journal*, 699, 1976, 702.

[47] E.g. J.H. REIMAN, *Privacy, Intimacy, and Personhood*, in *Philosophy & Public Affairs*, 6, 1976.

[48] For an insight in various approaches *see* M. EVERETT, *Can You Keep a (Genetic) Secret? The Genetic Privacy Movement*, in *Journal of Genetic Counseling*, 13, 4, 2004.

[49] A. WESTIN, *Social and political dimensions of privacy*, in *J Soc Issues*, 59, 2, 2003.

[50] A.L. ALLEN, *Genetic Privacy: Emerging Concepts and Values*, in M.A. ROTHSTEIN (ed.) *Genetic secrets: protecting privacy and confidentiality in the genetic era*, New Haven, 1997, 31, 33 (ed., 1997).

[51] G. LAURIE, *Genetic Privacy: A Challenge to Medico-Legal Norms,* cit., 67-68.

[52] G. LAURIE, *Genetic Privacy: A Challenge to Medico-Legal Norms,* cit., 6.

[53] *See* also G. LAURIE, *Privacy and the right not to know: a plea for conceptual clarity*, in R. CHADWICK, M. LEVITT, D. SHICKLE (eds.) *The Right to Know and the Right Not to Know: Genetic Privacy and Responsibility*, Cambridge, 2014, 41.

[54] G. LAURIE, *Genetic Privacy: A Challenge to Medico-Legal Norms,* cit., 182.

is subsumed under privacy, and when and how it is an effective means of safeguarding the interests of an individual, has been the subject of discussion.[55] For the purposes of further analysis, as a starting point it suffices to note that autonomy is an important aspect of genetic privacy, which can be further considered in terms of an informational and spatial state of separateness, disregarding its conceptual location. However, it neither is nor should be an absolute prerogative.[56] Thus, while it can be viewed as a way of control over the two privacy dimensions put forward by Laurie, one should not exclude other types of acceptable access avenues in the state of separation.

2.4. Legal foundations

In parallel to the advances in the area of genomics, there have also been advances in legal regulations in the field. In the European regional legal fora, genetic privacy is protected by legal instruments of both the Council of Europe and the EU. Some of the central instruments relevant for further discussion are presented below.

The Council of Europe has taken a rather overarching and often comprehensive approach to addressing genetic privacy in different contexts. To illustrate, the European Court of Human Rights (ECtHR) has anchored several facets of genetic privacy in the protection of private life under Article 8 of the Convention for the Protection of Human Rights and Fundamental Freedoms (ECHR). For example, in 2006 in an admissibility decision, the ECtHR pointed out that, "given the use to which cellular material in particular could conceivably be put in the future, the systematic retention of that material goes beyond the scope of neutral identifying features such as fingerprints, and is sufficiently intrusive to constitute an interference with the right to respect for private life set out in Article 8 § 1 of the Convention".[57] In *S. and Marper v. the United Kingdom* it found that retention of cellular samples per se is interference with the right to private life.[58] Acknowledging the identification capacity DNA profiles have, retention of such profiles was also considered to be an interference with the right to private life.[59] Any interference with the right to private life could be justified in accordance with Article 8(2) of the ECHR.

Under this very same provision, the ECtHR has laid down foundations for the protection of integrity and informed consent as a tool in that regard.[60] The Convention also protects several other rights that can be triggered by an integrated genetic enterprise, as discussed previously, including freedom of expression under Article 10 ECHR. Moreover, under the auspices of the Council of Europe further instruments have been adopted with a different focus, a different degree of detail and a different legal force relevant for regulating medical care or research generally, or genetic interventions specifically. Examples of these instruments include the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine (Biomedicine Convention) which aims to protect the dignity and iden-

[55] *See* R. ANDORNO, *The right not to know: an autonomy based approach*, in *Journal of Medical Ethics*, 30, 2004.
[56] G. LAURIE, *Genetic Privacy: A Challenge to Medico-Legal Norms,* cit., 183-185.
[57] *Van der Velden v. the Netherlands* (Application no. 29514/05, 7 December 2006), 8.
[58] *S and Marper v. United Kingdom*, *op. cit.*, para. 73.
[59] *S and Marper v. United Kingdom*, *op. cit.*, para. 75.
[60] *See* e.g. *Y.F. v. Turkey* (Application no. 24209/94, 22 July 2003), para. 33.

tity of all human beings in the application of biology and medicine,[61] and its additional protocols on genetic testing[62] and biomedical research.[63] They also include the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data,[64] as well as various recommendations, for example, Recommendation on the protection of health-related data,[65] Recommendation on the processing of personal health-related data for insurance purposes, including data resulting from genetic tests,[66] and Recommendation on research on biological materials of human origin.[67]

Within the EU, the approach to safeguarding genetic privacy is directed by the principle of conferral and how the EU has exercised these competences. Substantively, the question of genetic analysis triggers the application of the in vitro diagnostic medical devices (IVDMD) framework. Currently, the area is regulated under the IVDMD Directive which has been transposed in the national laws of the Member States and encourages application of the principles of the Biomedicine Convention.[68] However, from 26 May 2022 an IVDMD Regulation is expected to apply.[69] In regard to integrity protection, it refers to the principles set out in the Charter of Fundamental Rights of the European Union (CFREU),[70] and when genetic testing is offered in healthcare for health-related purposes it requires the Member States to ensure that information and counselling is provided. Implementing greater protective provisions and informed consent can be done nationally.[71] Genetic data protection is regulated under the GDPR.[72] When the EU law applies, so too does the CFREU.[73] Article 7 of the CFREU sets forth the right to private life generally, whereas Article 8 CFREU addresses data protection specifically. Moreover, the CFREU sets forth protection of a number of other rights, including freedom of expression and information as guaranteed by Article 11, freedom of the arts and sciences as guaranteed by Article 13, and the requirement for informed consent under Article 3. Generally, in accord-

[61] Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, ETS 164, Article 1.

[62] Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes, ETS No. 203.

[63] Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research, ETS No.195.

[64] Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data, ETS 108. See note 7 and 8 above regarding revision of this treaty.

[65] Recommendation CM/Rec(2019)2 of the Committee of Ministers to member States on the protection of health-related data.

[66] Council of Europe, Committee of Ministers, Recommendation CM/Rec (2016)8 on the processing of personal health-related data for insurance purposes, including data resulting from genetic tests.

[67] CoE Recommendation CM/Rec(2016)6 of the Committee of Ministers to member States on research on biological materials of human origin.

[68] Directive 98/79/EC of the European Parliament and of the Council of 27 October 1998 on in vitro diagnostic medical devices OJ L 331, 7.12.1998, 1–37, recital 33.

[69] Regulation (EU) 2017/746 of the European Parliament and of the Council of 5 April 2017 on in vitro diagnostic medical devices and repealing Directive 98/79/EC and Commission Decision 2010/227/EU (Text with EEA relevance) OJ L 117, 5.5.2017, p. 176–332, Article 113.2, recital 89.

[70] IVDMD Regulation, *op. cit.*, recital 89.

[71] IVDMD Regulation, *op. cit.*, Article 4(4).

[72] GDPR, *op. cit.*

[73] Charter of Fundamental Rights of the European Union OJ C 326, 26.10.2012, 391–407, Article 51(1). Case C-617/10, *Åklagaren v Hans Åkerberg Fransson*, ECLI:EU:C:2013:105. Moreover, there is a complex interplay between the EU law and ECHR, which could shape interpretation and application of EU law.

ance with Article 52(1), limitations to these rights are possible if they are provided for by law and respect the essence of those rights and freedoms. They must also be necessary and genuinely meet objectives of general interest recognised by the EU or protect the rights and freedoms of others.
What can be derived from the above is that common to the regulatory approaches in both legal orders is a focus on both facets of genetic privacy elaborated by Laurie, namely, its spatial and informational dimension. Given the nature of the specific instruments as well mandates for respective legal orders, it can be noted that the Council of Europe legal instruments that address genetic privacy from the perspective of biomedical interventions generally or genetic analysis specifically are rather detailed. This can be contrasted with the EU's approach to indicating adherence to the specific requirements, leaving the modalities to the Member States.

3. Genetic privacy in context: genetic analysis

3.1. On spatial and informational privacy and genetic analysis

The protection of genetic privacy in terms of enabling intervention that leads to genetic data and information, and return of the results, can at times be rather simple and straightforward to safeguard but at others rather complex and challenging. It touches upon such questions as who is the one being tested and who is a decision-maker, what data and information are acquired as a result of testing, and how they are used, and who is the beneficiary of the afforded legal protection. As was elaborated in Section 2, while spatial privacy focuses on the person's physical and psychological space, informational privacy focuses on sharing information with others. Who these others are, and consequently whether the matter leaves the spatial privacy category and enters informational privacy, might not be straightforward to ascertain. Arguably, adult testing when the person concerned consents herself to the intervention is the most straightforward case, yet even this is not simple. Multiple challenges and uncertainties regarding genetic privacy protection emerge when consent to the intervention is given on behalf of the person (e.g. a child) or when the testing concerns an embryo or foetus. To capture the spectrum of different challenges, and thus also create a wider platform for examining the meaning of genetic privacy in the era of personal data protection, this section focuses on a selection of different contexts in which questions pertaining to genetic privacy in relation to genetic analysis emerges. In particular, it looks at genetic analysis and privacy at the pre-birth and post birth stages; in regard to the latter, it considers both situations – i.e. where the person being tested can and cannot consent.

3.2. Privacy and genetic analysis from the healthcare intervention regulation perspective

3.2.1 Non-reproductive genetic analysis and decision-making

When the decision-maker and the person being tested is one and the same person (in situations concerning a competent person), a number of questions falling in the domain of spatial and informational privacy protection can, at least on the surface, be governed relatively easily through informed consent. It is well-established that informed consent is a means to control one's integrity, ensure respect

for the patient's integrity and enhance dignity in a medical context.[74] As set out in Article 5 of the Biomedicine Convention, a genetic intervention may only be carried out after the person concerned has given free and informed consent to it. Prior to giving consent, the person concerned shall receive appropriate information as to the purpose and nature of the particular genetic analysis being carried out as well as its consequences and risks. A similar requirement is also set forth in the Additional Protocol on Genetic Testing,[75] which also requires genetic counselling when testing concerns monogenetic disease, detecting a genetic predisposition or genetic susceptibility to a disease, or identifying the subject as a healthy carrier of a gene responsible for a disease.[76] In a similar way, IVDMD Regulation also sets forth a requirement of information, and directs the national law-makers to address the matter of informed consent,[77] whereas its predecessor, the IVDMD Directive hints at necessity to comply with the requirements of the Biomedicine Convention.[78] However, a closer look reveals limitations in using informed consent as a tool for safeguarding genetic privacy.

By way of illustration, the following can be noted. First, informed consent focuses on the intervention and associated consequences and risks (implications and results), be they directly associated with the analysis or related to the analysis. Both conceptually and legally, it primarily captures the relationship between the one offering a test, e.g. a care giver, and the one being tested, and not the relationship between the care-giver (or institution where the care-giver works) and the testing service provider. The extent to which genetic privacy intrusions occur outside such a relationship risks falling beyond the reach of this tool. This could create an expectation that privacy is safeguarded in the subsequent actions necessary for testing. However, this protection can be addressed in a different way, e.g. through different claims for establishing mechanisms that uphold privacy protection in subsequent steps, e.g. such as data protection regulatory requirements.

When the one giving consent and the one being tested is not the same person (persons lacking decision-making capacity), multiple challenges emerge, both in terms of authorising the intervention and returning the analysis result, in addition to other genetic privacy challenges, such as those illustrated above. Article 6 of the Biomedicine Convention and Chapter V of the Additional Protocol on Genetic Testing set the limitations regarding the permissible scope of intervention in such situations. First, the intrusion in genetic privacy is conditioned on there being a "direct benefit" for the person being

[74] The doctrine of informed consent is central to the legal and ethical regulation of biomedical interventions. Informed consent has been labelled "the modern clinical ritual of trust". P. R. WOLPE, *The Triumph of Autonomy in American Bioethics: A Sociological View*, in R. DEVRIES, J. SUBEDI (eds.), *Bioethics and society: Constructing the ethical enterprise*, Englewood Cliffs, 1998, 38-59. It has also been portrayed as means to ensure that a patient has not been deceived or coerced into a particular intervention. O. O'NEILL, *Some Limits of Informed Consent*, in *Journal of Medical Ethics*, 29, 2003, 4, 5. As a tool, it aspires to enhance self-determination and give patients greater control over their bodily (or other type of) integrity. It is well established that a medical intervention, even if it is of minor importance, constitutes an interference with the right to privacy. *Y.F. v. Turkey* (Application no. 24209/94, 22 July 2003) para. 33. The ECtHR has characterised a right to self-determination as an inalienable right. *See Plesó v. Hungary* (Application no. 41242/08, 2 October 2012), para. 66.

[75] Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes, ETS 203, Articles 8.1 and 9.1.

[76] Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes, ETS 203, Article 9.2.

[77] IVDMD Regulation, *op. cit.*, Article 4.

[78] IVDMD Directive, *op. cit.*, recital 33.

tested. Second, it is conditioned on the authorisation given by the person legally entitled to represent the person being tested. Finally, in the case of a minor, the testing should be deferred until the attainment of capacity to decide unless the delay is detrimental to the child's health or well-being.[79] Moreover, the minor's opinion should be taken into consideration in proportion to her age and degree of maturity, whereas (where the national law allows for decision-making on behalf of adults who do not have capacity to consent) the person concerned should, as far as possible, participate in the decision-making procedure.[80] Despite the layer of legal protections, safeguarding of the genetic privacy of minors has been of particular concern, especially in commercial genomic services.[81] Thus far, focused national regulatory responses remain scarce. For example, Norway has been working towards a law preventing parents from consenting to medically unmotivated genetic tests on behalf of their children.[82] In Sweden, in contrast, such a legislation does not exist and the matter is regulated under medical law and/or family law rules depending on the context and type of testing carried out.[83]

When informed consent (or assent) is given on behalf of someone, it serves as a means to enable an intervention, and thus access to a spatial state of separateness. When and in so far as the information is returned to the person consenting or others, the matter relates to an intervention in an informational state of separateness. That is directed by the principle of direct benefit under the reviewed legal instruments, which along with relevant modalities of a particular test shape the conditions for accessing the state of separateness. While the right to privacy in regard to genetic information applies, disclosures could be made in order to safeguard the health and well-being of the person concerned.[84] Moreover, interventions to safeguard the interests of others could be made.[85]

3.2.2 Reproductive genetic analysis and decision-making

In a reproductive context, one can draw a distinction between genetic analysis of the prospective parent or parents (e.g. preconception carrier genetic testing) and testing that is done at a later stage of the reproductive journey, either in the course of *in vitro* fertilisation or as part of antenatal care, both coupled with adequate genetic care services, in particular relevant counseling. The former rais-

[79] Additional Protocol on Genetic Testing, *op. cit.*, Article 10.

[80] Additional Protocol on Genetic Testing sets further safeguards for the protection of a minor's privacy. In accordance with Article 10, where according to law, a minor does not have the capacity to consent, a genetic test on this person shall be deferred until attainment of such capacity unless the delay is incompatible with the child's interests.

[81] H. C. HOWARD, D. AVARD, P. BORRY, *Are the Kids Really All Right? Direct-to-Consumer Genetic Testing in Children: Are Company Policies Clashing with Professional Norms?,* in *European Journal of Human Genetics*, 19, 2011, 1122. L. F. ROSS, H. M. SAAL, K. L. DAVID, R. R. ANDERSON, American Academy of Pediatrics, American College of Medical Genetics and Genomics, *Technical Report: Ethical and Policy Issues in Genetic Testing and Screening of Children*, in *Genetics in Medicine,* 15, 2013, 234.

[82] BIOTEKNOLOGIRÅDET, DNA-tester av barn utenfor helsevesenet (2018), www.bioteknologiradet.no/filarkiv/2018/07/2018-07-13-DNA-testing-av-barn.pdf (last visited 16/02/2021).

[83] *See* S. SLOKENBERGA *The standard of care and implications for paediatric decision-making: the Swedish viewpoint,* in C. Ó. NÉILL, C. FOSTER, J. HERRING, J. TINGLE (eds.) *Routledge Handbook of Global Health Rights,* Abingdon, 2021, 8.

[84] *See* Additional Protocol on Genetic Testing, *op. cit*., Articles 10 and 11.

[85] Additional Protocol on Genetic Testing, *op. cit*., Article 13.

es a number of ethical questions. However, from the perspective of genetic privacy in the course of medical care it is largely similar to the types of genetic analysis discussed above. The latter, interventions in the context of *in vitro* fertilisation and antenatal care, is of particular interest here. Central to all of these types of interventions is access to information about a future child at a time when, at least from the human rights perspective, this future child might not necessarily benefit from such protection.

To begin with, in the course of *in vitro* fertilisation, preimplantation genetic diagnostics – more accurately called preimplantation genetic testing or screening – can be carried out. This type of analysis is the only one that enables obtaining information prior to implantation, and can thus prevent a pregnancy termination or the passing of a particular mutation gene to an offspring.[86] In order to carry it out access to DNA from either gametes or embryos within 6 days of conception is required.[87] This can be done using various methodologies,[88] and be applied in different contexts. Acknowledging that they raise considerable ethical, social and legal questions from the privacy perspective, if approached as a state of separateness and one that is attributable to the respective subjects, the degree of intervention in genetic privacy can vary. For example, the analysis can have a restricted scope (e.g. a particular mutation) or it can aim at sequencing the whole embryonic genome.[89] It can also be applied in different contexts, such as cancer and HLA tissue typing,[90] and be relevant for speculative non-clinical applications, e.g. testing for the perfect pitch or intelligence.[91]

There are many types of non-invasive prenatal screening tests. They generally include several interventions (e.g. measurement of multiple analytes in the maternal serum, ultrasound investigation of the foetal development) and they often have poor accuracy.[92] More conclusive diagnostic analysis has traditionally required obtainment of a foetal biological material, through amniocentesis, chorionic villus sampling or fetal blood sampling.[93] Generally, techniques used in the analysis include cytogenetics to screen for fetal chromosomal anomalies or molecular genetic techniques that are generally applied for the purpose of identifying single-gene disorders.[94] While such interventions are generally highly accurate, they carry the risk of postprocedure foetal loss.

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[86] J. L. Simpson, A. Kuliev, S. Rechitsky, *Overview of Preimplantation Genetic Diagnosis (PGD): Historical Perspective and Future Direction*, in B. Levy (ed), *Prenatal Diagnosis*, New York, 2019, 23-43.

[87] J. L. Simpson, A. Kuliev, S. Rechitsky, *op. cit*.

[88] S. Chen, X. Yin, S. Zhang, J. Xia, P. Liu, P. Xie, H. Yan, X. Liang, J. Zhang, Y. Chen, H. Fei, L. Zhang, Y. Hu, H. Jiang, G. Lin, F. Chen, C. Xu, *Comprehensive Preimplantation Genetic Testing by Massively Parallel Sequencing*, in *Human Reproduction*, 36, 2021, 236.

[89] A. Kumar, A. Ryan, J. O. Kitzman, N. Wemmer, M. W. Snyder, S. Sigurjonsson, C. Lee, M. Banjevic, P. W. Zarutskie, A. P. Lewis, J. Shendure, M. Rabinowitz, *Whole Genome Prediction for Preimplantation Genetic Diagnosis*, in *Genome Medicine*, 7, 2015, 35. R. Winand, K. Hens, W. Dondorp, G. de Wert, Y. Moreau, J. R. Vermeesch, I. Liebaers, J. Aerts, *In Vitro Screening of Embryos by Whole-Genome Sequencing: Now, in the Future or Never?*, in *Human Reproduction*, 29, 2014, 842.

[90] J. L. Simpson, A. Kuliev, S. Rechitsky, *op. cit*.

[91] J. A. Robertson, *Extending Preimplantation Genetic Diagnosis: Medical and Non-Medical Uses*, in *Journal of Medical Ethics*, 29, 2003, 213.

[92] O. Cogulu, *Next Generation Sequencing as a Tool for Noninvasive Prenatal Tests,* in U. Demkow, R. Ploski (eds.), *Clinical Applications for Next-Generation Sequencing*. London, 2015, 173.

[93] O. Cogulu, *op. cit.*, 173.

[94] O. Cogulu, *op. cit.*, 173.

Non-invasive parental testing (NIPT) allows for analysis of a foetal DNA in a maternal blood sample. It is based on the analysis of cell free foetal DNA, which are fragments of DNA released into the pregnant woman's bloodstream[95] that is already present in the gestational mother's blood early in the pregnancy,[96] using, for example, advanced genomic technologies.[97] In 2011, when NIPT became clinically available, it was offered for selected foetal trisomies. It is now also used in other contexts, such as gender detection, and has the potential to expand to other applications.[98] There are different techniques that can be used for that purpose, for example, single-nucleotide polymorphism and whole-genome sequencing,[99] and they lead to different degrees of intervention into genetic privacy. It has already been demonstrated that it is possible to decipher the entire foetal genome from placental cfDNA. As the cost of the technology comes down, it is expected that the applications will expand to a wider range of medical conditions as well as behavioral traits (e.g. intelligence and aggression), although the accuracy of such interventions could be an issue.[100]

The focus of reproductive genetic testing lies predominantly, if not almost entirely, on the individual reproductive choice of the prospective mother or jointly with the prospective parents, e.g. in the case of use of preimplantation genetic analysis. In the course of antenatal care, intervention is carried out through the pregnant woman, and consequently, it is generally her consent to an intervention that governs the intervention, as well as her decision on the intervention of the foetal state of separateness. In both cases, the limits regarding the degree of permissible intervention prescribed by the applicable legal frameworks could apply. It is not precluded that the other prospective parent could be involved in medical care, e.g. by means of information and counselling. In a similar way, returned information will focus on the health status of the prospective child, and possibly be related to the health of the woman or both of the parents. In the event of pregnancy, the prospective mother's spatial privacy is used to access information about the foetus, and return of this information is subjected to the very same privacy provisions.

Governance of the state of separateness at a pre-birth stage from a human rights perspective is not a straightforward matter. It is well-established that matters relating to the continuation of pregnancy fall within the scope of private life and autonomy as protected under Article 8 ECHR.[101] Likewise, the use of assisted reproductive technologies generally, as well as the question of genetic analysis in that regard, attracts the protection of this article.[102] The ECtHR has stated that, "[i]n matters of general policy, on which opinions within a democratic society may reasonably differ widely, the role of the

[95] O. COGULU, *op. cit.*, 174.

[96] O. COGULU, *op. cit.*, 173-6.

[97] NIPT include several options: NGS of ccfDNA, PCR-based methods, microarrays and single foetal cell genome analysis. O. COGULU, *op. cit.*, 176.

[98] *See*, for example, Cho suggesting the potential in regard to other chromosomal imbalances, E. CHO, *Whole Genome Sequencing Based Noninvasive Prenatal Test*, in *Journal of Genetic Medicine,* 12, 2005, 65.

[99] D. MUZZEY, *The Technology and Bioinformatics of Cell-Free DNA-Based NIPT,* in L. PAGE-CHRISTIAENS, H. KLEIN (eds.), *Noninvasive Prenatal Testing (NIPT),* Cambridge, 2018, ch.3.

[100] *See* Nuffield Council on Bioethics, *Non-invasive prenatal testing: ethical issues*, 2017. https://www.nuffieldbioethics.org/wp-content/uploads/NIPT-ethical-issues-full-report.pdf (last visited 10/02/2021) paras. 1.32 – 1.34.

[101] *A.K. v. Latvia* (Application no. 33011/08, 24 June 2014), para. 63.

[102] *Costa and Pavan v. Italy* (Application no. 54270/10, 28 August 2012), para.57.

domestic policy-maker should be given special weight."[103] In *S.H. and Others v. Austria* the Court emphasised the sensitive moral and ethical issues in fast-moving medical and scientific developments, and afforded states wide margin of appreciation.[104] In the case of *Costa and Pavan v. Italy*, the Court acknowledged that access and use of preimplantation genetic diagnosis raises sensitive amoral and ethical questions,[105] but it refrained from reflecting on margin of appreciation in regulating the technology generally, and indicated that medical and scientific developments could have a bearing on the margin of appreciation.[106] Access to genetic information at a pre-birth stage, while undoubtedly relating to enabling informed reproductive choices, also triggers a number of other important interests as well as revealing genetic information of the unborn. While one could argue that access to foetal genetic information could be a matter on which states generally enjoy a margin of appreciation, how wide this margin of appreciation is in regard to the technology generally, as well as a spatial and informational state of separateness, could depend on a number of factors, including the context in which the information is accessed and the medical and scientific state of art on the matter.

Article 12 of the Biomedicine Convention specifically regulates predictive genetic tests. It enables tests to be performed that are "predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease, and may be performed only for health purposes." Although it focuses on the health purposes for an individual, and so one could question its effects on prenatal testing, the explanatory report to the Convention does point out that this provision is not intended to place "any limitation on the right to carry out diagnostic interventions at the embryonic stage to find out whether an embryo carries hereditary traits that will lead to serious diseases in the future child."[107] The Additional Protocol on Genetic Testing *expressis verbis* excludes from the scope of application genetic tests that are "carried out on the human embryo or foetus." Although that is not mentioned expressly in Article 2(2) of the Additional Protocol on Genetic Testing, NIPT is also intended to be exempted from the scope of application of the Protocol.[108] It could, however, be further regulated by the respective instruments of the contracting parties.

Thus, based on the current state of the law, one can draw a preliminary conclusion that there are increasingly sophisticated methods with which to access increasing amounts of genetic information at different points within the pre-birth stage. However, the legal protection – at least in relation to genetic analysis as a biomedical intervention – is rather weak and one can question whether the existing mechanisms are fit to deal with the challenge of enhanced access to the genome at a pre-birth stage, and implications that this could bring along. There are a considerable number of reasons for why the reviewed instruments are formed in a particular way and for the ECtHR's caution in the field.

[103] *Maurice v. France* (Application no. 11810/03, 6 October 2005). para. 117.
[104] *S.H. and Others v. Austria* (Application no. 57813/00, 3 November 2011), para. 97.
[105] Costa and Pavan v. Italy, *op. cit*., para. 58.
[106] Costa and Pavan v. Italy, *op. cit*., para. 67.
[107] Explanatory Report to the Convention for the protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, para. 83.
[108] Explanatory Report to the Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes, para.31.

Any question relating to the conception and continuation of a pregnancy, and the application of the existing technologies, raises considerable questions and it is difficult to reconcile the various views. While it might be of minor importance to further consider the state of separateness of those which will never be born, for those who are born testing at a pre-birth stage opens up an unlocked informational privacy. It raises further questions about whether and how interventions should be regulated, whether the existing approaches are adequate, and whether mechanisms are or should be in place that informed of interventions in the state of separateness at a pre-birth stage.

3.3. Managing genetic information under the healthcare framework

Article 10 of the Biomedicine Convention addresses the right to private life and information. It affirms that everyone has the right to respect for private life in relation to information about her health. While everyone is entitled to know any information collected about her health, the wishes of individuals not to be so informed should also be observed.[109] In the context of genetic privacy this means, for example, accommodating the wish not to know about particular genetic risks. Those falling within the scope of "everyone" remains to be prescribed by the parties of the Convention. However, the wording of Article 10 does not seem to preclude its application at a post-birth stage for pre-birth interventions.[110]

In addition to the specific purpose of genetic analysis and information processed in that regard, the issue of incidental findings and how they are managed is also of concern. Although one can draw a distinction between the results of a deliberate search, e.g. a list of pathogenic and likely pathogenic variants that laboratories search for in specified genes, and the more common use of the term incidental findings, namely, when genetic findings are discovered unexpectedly, both could be managed under the informed consent requirement. However, depending on what consent is being given to, how the capacity to deliver incidental findings changes in a particular situation, and how the right to information is exercised,[111] the contours of one's genetic privacy might be affected.

In addition to the person being tested, her relatives could also have an interest in the risk information. Different approaches to addressing the familial nature of genetic information have been put forward, such as a personal account model that focuses on patient choice and confidentiality, and a joint account model that focuses on the reasons for not disclosing information to family members.[112] Although privacy is a qualified right, and as such, it allows for restrictions when there are overriding reasons, the Additional Protocol on Genetic Testing does not provide conclusive answers. It mandates that the person being tested is informed that the obtained information can be relevant to the health of other family members, and that this is duly addressed in the information to the person.[113] The communication of this information is left for the individual states. Thus, it could well be that an individual's information is shared with her relatives, even though she would prefer it not to. Although the United Kingdom is not part of the Additional Protocol on Genetic Testing, it is relevant to high-

[109] Biomedicine Convention, *op. cit.*, Article 10.
[110] Explanatory Report to the Biomedicine Convention, *op. cit*., para.18.
[111] Biomedicine Convention, *op. cit.*, Article 10.
[112] M. PARKER, A. M. LUCASSEN, *Genetic Information: A Joint Account*?, in *BMJ*, 329, 2004, 165.
[113] Additional Protocol on Genetic Testing, *op. cit.*, Article 18.

light a case from England and Wales in 2020 that acknowledged the acceptability of breach of confidentiality in the context of genetic information in specific circumstances. In *ABC v St Georges Health Trust*, the High Court of England and Wales[114] decided that a balance needed to be struck between disclosure of a disease for the benefit of a relative and the wishes of the patient. It could be, as in this specific case, that the interests of a relative will prevail, which justifies intervention with the proband's state of separateness.

3.4. Privacy and genetic analysis under the GDPR

The GDPR is clear that its focus is the protection of personal data of natural persons.[115] However, it cannot be precluded that the GDPR may also cover data obtained pre-birth about a natural person once the criteria of a "data subject" are satisfied (post birth). The GDPR will apply to the processing of health and genetic data in that regard,[116] including when the intended genetic analysis is carried out, on condition that the material protection of the GDPR is triggered, e.g. medical care is provided by a care provider in the EU.[117] However, the processing itself will not necessarily require the consent of a data subject.[118] Generally, it will be enough that medical care is lawfully provided in order for the genetic data to be lawfully processed under Articles 6(1) and 9(2) GDPR. That is to say, a lawful medical intervention can set the foundation for the subsequent steps needed to complete the respective medical intervention, thus enabling more sophisticated interventions in an individual's state of separateness. What combination of grounds set out in Article 6(1) and 9(2) GDPR will be triggered depends on the circumstances, including whether consent is given by the person being tested or on behalf of that person, as well as relevant regulations at the national level.

Disregarding whether the consent to genetic testing as a biomedical intervention is given by the person being tested or on behalf of this person, the consent or authorisation for the genetic intervention is subject to withdrawal.[119] One can question the effects of such a withdrawal from a genetic privacy perspective. If the consent is perceived in terms of a medical relationship in which the care provider and recipient are the focus, it ends this relationship in the respective part. Whether and to what extent it could stop further occurrences, such as testing in a laboratory, depends on multiple considerations. Withdrawal of consent in the broader genetic analysis context would mean that not only the person concerned wishes to shield herself from the intervention through, for example, return of the results, but also from analysing the provided sample. Whether and to what extent that could be possible in a particular case depends on multiple practical and legal considerations, including how the

[114] ABC v St George's Healthcare NHS Trust, South West London and St George's Mental Health NHS Trust and Sussex Partnership NHS Foundation Trust.

[115] GDPR, *op. cit.*, Article 1.1.

[116] *See* GDPR, *op. cit.*, Article 2, material scope.

[117] *See* GDPR, *op. cit.*, Article 3, territorial scope.

[118] For how the combination of Articles 6 and 9 plays out in healthcare in the EU under the GDPR *see* DG Health and Food Safety, Assessment of the EU Member States' rules on health data in the light of GDPR, Luxembourg: Publications Office of the European Union, 2021. www.ec.europa.eu/health/sites/health/files/ehealth/docs/ms_rules_health-data_en.pdf (last visited 12/02/2021), 28-30.

[119] Biomedicine Convention, *op. cit.*, Articles 5 and 6. Additional Protocol on Genetic Testing, *op. cit.*, Articles 9.2 and 12.4.

provision of the laboratory services is organised, the legal grounds for the laboratory to process the sample (e.g. in-house or third party and thus a separate legal relationship), as well as how efficiently the withdrawal can practically be communicated. While full withdrawal might not necessarily always be possible, that is something that could, and in fact one could argue that should, be communicated as part of the informed consent process to genetic testing as an intervention in the biology and medicine generally or healthcare specifically.

In addition to shaping the lawfulness of a particular intervention, the GDPR sets out a number of detailed obligations on the controllers and processors and establishes the rights of the data subjects.[120] Even if the consent to medical care triggers lawfulness of analysis, and subsequent analysis is beyond the control of the data subject from the healthcare regulatory perspective, the aim of the detailed data protection requirements and provision of such rights as the right to information to the data subjects is to establish a mechanism that protects personal data in different flows and places the individual, at least theoretically, in the controlling position. These mechanisms suggest measures to safeguard the state of separateness in accordance with the prescribed requirements and limitations.

4. Genetic privacy in context: scientific research and publication

4.1. On genomic data, scientific research and publication

Advances in science commonly emanate from questioning the status quo of a particular field and expanding its borders, and then establishing new or enhanced applications.[121] That requires not only carrying out scientific research that challenges the limitations of the existing knowledge but also effectively disseminating the existing knowledge and engaging in rigorous scientific debate in line with the applicable standards.[122]

In the last decades, increasing emphasis has been placed on enhancing data sharing and on the openness of science and its democratisation. As scientific research has enjoyed increased attention in the human rights arena,[123] it has become common to treat openness not only as a good research practice but also as a means to fulfil the obligations stemming from Article 27(1) UHDR and Articles 15(1)(b) and 15(3) ICESCR. In the field of genomics, a robust culture of data sharing has been developed.[124] Genomic data sharing can take many forms. Byrd et al. have distinguished between the fol-

[120] *See* GDPR, chapters II, III and IV.

[121] On reflections of the tasks of science generally and correction of errors specifically *see* A. W. BROWN, K. A. KAISER, D. B. ALLISON, *Issues with Data and Analyses: Errors, Underlying Themes, and Potential Solutions*, in *Proceedings of the National Academy of Sciences*, 115, 2008, 2563.

[122] *See* e.g. duty to share knowledge under Article 15 of the Universal Declaration on Bioethics and Human Rights and obligation to publish research in line with Article 28.3 of the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research, ETS 195. *See* also A. K. BEFRING, *Kunstig Intelligens Og Rettslige Perspektiver,* in A. K. BEFRING, I. J. SAND (eds.), *Kunstig intelligens og big data i helsesektoren*. *Rettslige perspektiver*, Gyldendal, 2020.

[123] *See*, e.g. Committee on Economic, Social and Cultural Rights, *General comment No. 25 (2020) on science and economic, social and cultural rights (article 15 (1) (b), (2), (3) and (4) of the International Covenant on Economic, Social and Cultural Rights)*, 30 April 2020, E/C.12/GC/25.

[124] J. B. BYRD, A. C. GREENE, D. V. PRASAD, X. JIANG, CASEY S. GREENE, *Responsible, Practical Genomic Data Sharing That Accelerates Research*, in *Nature Reviews Genetics*, 21, 2020, 615.

lowing four: public data sharing, controlled-access sharing, clique sharing and sharing upon request, with each of these models possessing different sharing structures, levels of difficulty and variable levels of risk associated with sharing.[125] Additionally, initiatives to improve openness have been established and avenues for bottom-up sharing have been created,[126] as have tips for open sharing of genetic data, though with the caveat that they are only relevant when data sharing restrictions do not apply.[127] However, the increasing amounts of available genomic data facilitate establishing linkages between phenotype and genotype datasets, which can ultimately serve as grounds for re-identification of the research participants,[128] thus raising concerns about the layer of privacy over the data subject.

4.2. Scientific research

Tissue samples that have been removed for the purposes of or in the course of a medical intervention could be retained for other purposes if that is done in line with appropriate information and consent procedures.[129] It has been acknowledged that what is appropriate could vary in different circumstances and both opt-in and opt-out mechanisms could generally be compatible with this requirement.[130] When the retained material is used for scientific research, further requirements are of relevance. Within the Council of Europe, detailed requirements addressing genetic privacy in the context of scientific research are set forth in Recommendation of the Committee of Ministers to Member States on research on biological materials of human origin.[131] While this recommendation has a broad scope of application and it captures further use of biological material that has been collected for other purposes, as in the context of this article, i.e. genetic analysis, it expressly excludes from the scope of application embryonic and foetal material.[132] In regard to further use of biological materials collected in the course of an intervention, unless the intended research is covered by the previously obtained consent, it suggests that consent should be sought and reasonable efforts to contact the person should be made, whilst also respecting the wish of the person not to be contacted.[133] Only exceptionally, where the attempt to contact the person proves unsuccessful, can research be carried out subject to further conditions.[134] Further requirements apply to research involving biological material from persons unable to consent, thus establishing rather narrow situations for fur-

[125] J. B. Byrd, A. C. Greene, D. V. Prasad, X. Jiang, Casey S. Greene, *op. cit.*
[126] A. Middleton, *Your DNA, Your Say*, in *New Bioeth*., 23, 1, 2017, 74.
[127] A. V. Brown, J. D. Campbell, T. Assefa, D. Grant, R. T. Nelson, N. T. Weeks, S. B. Cannon, *Ten Quick Tips for Sharing Open Genomic Data*, in *PLOS Computational Biology*, 14, 2018, e1006472.
[128] G. Gürsoy, F. C. P. Navarro, M. Gerstein, *FANCY: Fast Estimation of Privacy Risk in Functional Genomics Data*, in *Bioinformatics*, 36, 2020, 5145.
[129] Biomedicine Convention, *op. cit*., Article 22.
[130] Explanatory Report to the Biomedicine Convention, *op. cit*., paras. 135-138, in particular para. 137. Genetic tests for research purposes are excluded from the scope of Additional Protocol on Genetic Testing, *see* Article 2(2)(b).
[131] Recommendation CM/Rec(2016)6 of the Committee of Ministers to member States on research on biological materials of human origin.
[132] Recommendation on research on biological materials of human origin, *op. cit.*, Article2(2).
[133] Recommendation on research on biological materials of human origin, *op. cit.*, Article 21(2)(a).
[134] Recommendation on research on biological materials of human origin, *op. cit.*, Article 21(2)(b).

ther use of the material.[135] As such, while the intention is for the individual to retain control over the further interventions in the context of research, there are also situations when it has been deemed legitimate to intervene in the absence of exercise of this control.

The GDPR applies to the processing of personal data, including genetic data in scientific research, and in that regard establishes a research-facilitating regime.[136] However, what exactly scientific research is within the meaning of "scientific research" under the GDPR, and thus benefiting from this regime, is still a subject of open discussion. It can be expected that the CJEU will eventually be called upon to provide answers that are more conclusive on this matter.[137] Some contours of the concept are sketched out in recital 159, which is the non-binding part of the legal act but also commonly used for ascertaining the aim of the legislator. It states that the notion of research GDPR "should be interpreted in a broad manner including, for example, technological development and demonstration, fundamental research, applied research and privately funded research". Research "should also include studies conducted in the public interest in the area of public health".[138] This broad take on an exception, which as a rule under EU law shall be interpreted narrowly, has been subject to interpretation considerations that risk undermining the approach that the legislator has taken.[139]

The scientific research regime could be said to rest on at least the following three interrelated pillars: data protection principles, data subject's rights and existence of adequate safeguards. Scientific research ticks the box of purpose limitation and storage limitation under Article 5 GDPR, albeit (from a literal interpretation of Article 6(1)) falling under the general rules on lawfulness of data processing, coupled with one of the requirements set out in Article 9(2) GDPR. Those enable lifting the ban on the processing of special categories of personal data. The different legal bases set out in Article 6(1) GDPR focus on demonstrating legitimate interest in the data subject's state of separateness, and their deployment might not be dependent on the wishes of the data subject. Put differently, storage for the purposes of scientific research and the research itself is a legitimate cause for further processing of personal data. Under the GDPR, informed consent is difficult to classify as a "right" in the same way as it is a "right" in the context of self-determination. While there is a right to withdraw consent under Article 7(3) GDPR, consent is only one of several possible legal bases for a lawful processing of the personal data under both Article 6(1) and 9(2) GDPR. There could be occasions where *informed consent* is used as a legal basis for data processing for scientific research. However, there could be many occasions when consent is irrelevant or not even an optimal possibility, such as when a research institution is a public body.[140] For example, in the case of public institutions, a legal obligation or task carried out in the public interest could be of particular relevance (Article 6(1)(c) or 6(1)(e) coupled with a research regulatory instrument at the EU or national level in line with Article 9(2)(j) GDPR). In the case of private institutions, though not without a challenge in the application of this

[135] Recommendation on research on biological materials of human origin, *op. cit.*, Article 21(5).
[136] C. STAUNTON, S. SLOKENBERGA, D. MASCALZONI, *op. cit.*, 1159.
[137] Consolidated version of the Treaty on European Union OJ C 326, 26.10.2012, 13–390, Article 19.1.
[138] GDPR, *op. cit.*, Recital 159.
[139] EUROPEAN DATA PROTECTION SUPERVISOR, *A Preliminary Opinion on data protection and scientific research*, 6 January 2020, 12.
[140] GDPR, *op. cit.*, recital 43.

provision, legitimate interest is safeguarded in Article 6(1)(f) (as guided by 6(4)), coupled with the mentioned research regulatory instrument at the EU or national level in line with Article 9(2)(j) GDPR. Although the data subject can have limited control as to whether data are used in scientific research, the GDPR can nonetheless be viewed as creating a certain level of personal data protection to the data subject through providing for several rights for the data subject. The rights the GDPR sets out as individual rights in Chapter III include the right to information, access rights, right to rectification, right to be forgotten, right to the restriction of processing, right to data portability and right to object. Other rights include the right to lodge a complaint and to compensation. The individual rights seek to ensure that the data subject is able to pursue a bottom-up control and it offers, at least under the law, a means to claim protection for the informational state of separateness warranted under the GDPR. However, there are several challenges in regard to safeguarding the state of separateness through the GDPR provisions. One occurs within scientific research in relation to the trade-off between individual rights as protected under Chapter III and adequate safeguards, where this trade-off is not dependent on the data subject but the factual circumstances in the case and modalities in the national law or EU law regulating scientific research.[141] Another is of a more generic nature and requires acknowledging the vulnerable status of the data subject: first, in relation to the controller (research institution), trusting that it will diligently complete the review, and second in relation to the data protection authority and its prioritisation. Fulfilment of these obligations is incentivised by several means, including responsibility and liability risks. However, to a considerable degree, it also requires that the data subject is proactive in protecting her own rights.

Overall, the data protection framework set up by the GDPR could be said to fill the state-of-separateness place left by the limitations of self-determination and the reach of consent. However, it is a different question how well that is done. While the GDPR was not the first to enter this field, it can surely be said to be more comprehensive and demanding than its counterparts. If data protection and by extension informational privacy is a value that should be upheld, then enhanced protection, even if it means adjustments within the scientific research community, is welcome. However, one can question to what extent the GDPR actually enhances privacy, which is understood here as a state of separateness. It clearly sets up a controlled access mechanism, yet it does not require the data subject to be in control. The control is assigned at the level of the GDPR, as well as with the EU legislature and national legislature, to various actors who could have an interest in entering the state of separateness. Only when circumstances so require, and even then often only at the initial stage, is control given to the data subject.

4.3. Scientific publication

Scientific publication is not only a means to further research and improve clinical practice[142] but also a means to safeguard freedom of expression and democratic society,[143] as well as protect the rights

[141] *See* S. SLOKENBERGA, *Setting the Foundations: Individual Rights, Public Interest, Scientific Research and Biobanking* in S. SLOKENBERGA, O. TZORTZATOU, J. REICHEL (eds), *op. cit.*, 16.

[142] A. ATTYÉ, *Data Sharing Improves Scientific Publication: Example of the "Hydrops Initiative"*, in *European Radiology*, 29, 2019, 1959.

and interests of research participants and the interests of society. One way of safeguarding these rights and interests is by emphasising the need to make research results public.[144] Scientific publication is a form of expression safeguarded under the right to freedom of expression set forth in the international human rights catalogues as well as in the EU legal order. Under the ECHR, for example, it is expressly addressed as a limited right,[145] and under the CFREU it is subject to limitations as set out in Article 52(1) CFREU. Scientific publication could be said to be the exact opposite of the protection of privacy. Instead of keeping information private, it focuses on exposing the necessary data for the public benefit. While the publication itself, as a printed or digitally presented material, might not necessarily require inclusion of personal data, the open science requirements, essential for furthering scientific research, have shaped best scientific publication practice and could require that.[146] Practices such as these in light of increasingly easily identifiability have raised a number of concerns relating to privacy and data protection.[147] While they are of importance in the context of genetic research, they are by no means exclusive to genetic data.[148]

The GDPR acknowledges that freedom of expression could conflict with the mechanism set up for the protection of personal data. As a general principle, it enables the Member States to reconcile freedom of expression for academic purposes with the GDPR. In that regard, in accordance with Article 85 the Member States are allowed to derogate from Chapter II (principles), Chapter III (rights of the data subject), Chapter IV (controller and processor), Chapter V (transfer of personal data to third countries or international organisations), Chapter VI (independent supervisory authorities), Chapter VII (cooperation and consistency) and Chapter IX (specific data processing situations) provided that these derogations are necessary to reconcile the right to the protection of personal data with freedom of expression and information. As rightly pointed out by the European Data Protection Supervisor, the scope of exemption exceeds that established under the research regime under Article 89

[143] *See* Parliamentary Assembly, Recommendation 1762 (2006) Academic freedom and university autonomy, para 14.

[144] Recommendation on research on biological materials of human origin, *op. cit.*, Article 23(2).

[145] *See* ECHR, *op. cit*., Article 10.

[146] *See* D. Mascalzoni, H. B. Bentzen, I. Budin-Ljøsne, L. A. Bygrave, J. Bell, E. S. Dove, C. Fuchsberger, K. Hveem, M. Th. Mayrhofer, V. Meraviglia, D. R. O'Brien, C. Pattaro, P. P. Pramstaller, V. Rakić, A. Rossini, M. Shabani, D. J. B. Svantesson, M. Tomasi, L. Ursin, M. Wjst, J. Kaye, *Are Requirements to Deposit Data in Research Repositories Compatible With the European Union's General Data Protection Regulation?,* in *Annals of Internal Medicine*, 170, 5, 2015, 332.

[147] E.g. EL Mmam et al. have demonstrated that the overall success rate for all re-identification attacks was approximately 26 and 34% for health data. K. El Emam, E. Jonker, L. Arbuckle, B. Malin, *A systematic review of re-identification attacks on health data*, in *PLoS One*, 6, 2011. Already in 2004 it was demonstrated that unique identification is possible with access to an individual's 75 single-nucleotide polymorphisms (SNPs). Z. Lin, A.B. Owen, R.B. Altman, *Genomic Research and Human Subject Privacy*, in *Science*, 305, 5681, 2004.

[148] S. Dyke, E. Dove, B. Knoppers, *Sharing health-related data: a privacy test?*, in *npj Genomic Med*, 1, 2016. However, whether danger associated re-identification in regard to genetic data in comparison with other types of data, is debated. For insights in the debates as well as argument that the dangers of re-identification for genetic and non-genetic data are rather similar, T.J. Kasperbauer, P. H. Schwartz, *Genetic Data Aren't So Special: Causes and Implications of Reidentification*, in *The Hastings Center Report*, 5/50, 2020.

GDPR.[149] Ultimately, the Member States have the power to create the necessary legal environment to remove unnecessary hindrances for disseminating the research.[150] In the context of genetic privacy, one of the central elements is this platform allows for silencing the data subject's interest in controlling its state of separateness, and from the perspective of the adequate legal basis, it adds on another step that could be taken with personal data without a data subject's control. In line with the general requirements stemming from the CFREU, any restriction needs to pass the proportionality test.[151] The validity of these exemptions, however, depends on notification to the European Commission.[152] The exact activities that fall under freedom of expression for the purposes of academic expression are not clearly defined, but they can be expected to be broadly interpreted to achieve the purpose of this objective.[153]

Given the close ties between the CFREU and ECHR, it should be borne in mind that the ECtHR has a well-established case law on safeguarding freedom of expression, which is a means of safeguarding a democratic society.[154] In the case of *Sorguç v. Turkey*, it affirmed the freedom to conduct research and distribute knowledge and truth without restriction set out in the Recommendation 1762 (2006) by the Parliamentary Assembly of the Council of Europe.[155] Generally, any exceptions to a right to freedom of expression need to be construed strictly.[156] The need for restrictions to this right emerge from a pressing social need and should be proportional to the aim pursued. Although the ECtHR has not had the opportunity to consider publication of genetic data, in the health data context it has provided some guidance for reconciling the interests at stake. In elaborating on the balance between freedom of expression and protection of private life, it noted that "a fundamental distinction needs to be made between reporting facts – even if controversial – capable of contributing to a debate in a democratic society and making tawdry allegations about an individual's private life."[157] At the same time, "the protection of personal data, not least medical data, is of fundamental importance to a person's enjoyment of his or her right to respect for private and family life as guaranteed by Article 8 of the Convention."[158] While the states could be afforded a certain margin of appreciation when deciding what "respect" for private life shall be ensured in particular circumstances, existence of a national law that balances the conflicting interests and provides protection is important.[159] Given the parallels between health data and genetic data, while acknowledging the important differences, and

[149] European Data Protection Supervisor, A Preliminary Opinion on data protection and scientific research, 6 January 2020. www.edps.europa.eu/sites/edp/files/publication/20-01-06_opinion_research_en.pdf (last visited 08/02/2021), 10.

[150] As emphasised in C-73/07, although the objective requires broad interpretation, the restrictions must apply only in so far as is strictly necessary. *See* C-73/07, Satakunnan Markkinapörssi and Satamedia, ECLI:EU:C:2008:727, para. 56.

[151] The limitations must not only be proportional, but also necessary and genuinely serve the purpose. *See* CFREU, *op. cit.*, Article 52.1.

[152] GDPR, *op. cit.*, Article 85.3.

[153] GDPR, *op. cit.*, recital 153.

[154] *See* e.g. *Handyside v. United Kingdom* (Application no. 5493/72), 7 December 1976, para. 49.

[155] *See Sorguç v. Turkey* (Application no. 17089/03) 23 June 2009, paras. 35 and 21.

[156] *Hertek v. Switxerland* (Application no. No. 53440/99) 17 January 2002, para.46.

[157] *Biriuk v. Lithuania* (Application no. 23373/03, 25 November 2008), para. 38.

[158] *Biriuk v. Lithuania*, *op. cit.*, para. 39.

[159] *Biriuk v. Lithuania*, *op. cit.*, paras. 44-46.

ISSN 2284-4503

considering the fragility of anonymity attributable to genetic data it could be expected that even more stringency is required. This, however, raises the question of evaluating the need for data sharing generally in order to shape such laws and policies, as well as the need for broader inquiries in the field.

5. Some reflections on genetic privacy in the era of data protection

This article has examined how the data protection requirements enshrined in the GDPR relate to shaping genetic privacy in the context of a complex and integrated enterprise of genetic testing. It began by setting the foundations for the inquiry. In that regard, it recapped some of the central scientific advances in the field and reflected on enhanced access to genetic information. It noted the different interests at stake in accessing genetic information and reviewed its conceptual and legal foundations. Thereafter, it looked at genetic analysis as a complex enterprise, examining genetic privacy in the context of genetic testing at a pre- and post-birth stage under the health and biomedical interventions legal frameworks and the GDPR, as well as scientific endeavours, through scientific research and publications. What is left to do now is to reflect on what these findings suggest about genetic privacy in the era of data protection.

There are rather stringent requirements relevant for genetic testing under health and biomedical care regulatory instruments. However, their strength in safeguarding genetic privacy stops with the limits attributed to the doctrines (and relevant legal mechanisms) of informed consent and scope of the respective legal instruments. One central limitation is safeguarding the state of separateness in the subsequent steps of genetic testing that follow the removal of a sample. Here, other complementary mechanisms that safeguard genetic privacy are necessary. Data protection regimes such as the GDPR can generate complementary protection effects. In terms of human rights and privacy protection, one could argue that the GDPR fulfils a state's positive duties and sets standards that protect privacy.

A particularly challenging situation emerges in relation to genetic analysis at a pre-birth stage. In this situation, intervention in a state of separateness is governed by the rights of the prospective mother or parents, depending on the exact context. As is clear from the health and biomedical care regulatory instruments, this type of testing is left for the signatories of the Biomedicine Convention to regulate on through reconciling the various interests at stake and finding ways to address sensitive issues in a manner acceptable in a democratic society at the national level. Here, however, the GDPR does not generate a complementary effect until after the status of a data subject is retained. This has the potential to lead to a situation where the same information needs to be treated differently at different points in time. Such a de facto situation is not unique to the GDPR and can also be noted in regard to the use of human biological material for other purposes than initially tested for.[160] It can be noted, however, that the Council of Europe has taken steps to address the challenge concerning the stark difference in data protection from the moment the status of a data subject is obtained. It has done this through a Recommendation to its Member States and urged to ensure adequate protection

[160] *See* Recommendation on research on biological materials of human origin, *op. cit.*

to such data.[161] They could become relevant in so far as these data are attributable to the prospective parents, or once the prospective child has become a data subject within the meaning of the respective data protection instrument. That could, however, result in a situation where by the time of birth the notion of genetic privacy has turned into genetic transparency in some contexts.

One of the central points that emerges is the limited control that the data subject has over genetic data and their further processing for other legitimate purposes, in particular, scientific research and publication. The GDPR is designed in such a way that the party who has an interest in processing personal data has to demonstrate an adequate legal basis to do so and the ability to lift the processing ban for genetic data as a special category of data. Moreover, it has to comply with other applicable requirements. This approach is intended to prevent illegitimate interventions in the state of separateness. At the same time, it excludes some applications which could be relevant in open publication systems from the scope of application (e.g. personal use) where misuse of the processed data could be a concern. It is of particular importance that adequate mechanisms are put in place that address misuse of this information, such as effective discrimination protection prevention measures.[162] Moreover, if the individual's state of separateness is a value deemed worthy to safeguard, enhanced transparency regarding the genetic enterprise is needed. It should also be explored whether and how persons consenting to genetic analysis are adequately informed of the breadth and depth of possible intervention without their further control (e.g. scientific research and publication).

Unlike scientific research, scientific publication is not regulated through the complex scientific research regime under the GDPR unless one argues that scientific publication is a constituting element of scientific research. While the issue could be worth discussing further, here it suffices to note that the structure of the GDPR could be argued to speak against it. Thus, the specific requirements provided for in the GDPR addressing freedom of expression would be of relevance. While the scope of possible derogations is striking on the surface, one can question whether the GDPR can be faulted for privacy impacts? As was discussed in section 4, scientific research in the context of personal data triggers the protection of two human rights, both of which are of a non-absolute nature. States are placed in the position of having to find a balance between them by reconciling the different rights and interests at stake. From this perspective, one could argue that the GDPR affords all data protection standards to safeguarding personal data in the context of scientific publications, except when the Member States find it necessary to strike a different balance. For those cases, it provides a platform for this to be done. From this perspective, Article 85 GDPR has an awareness-raising function on the balancing of different rights, whilst also signalling the EU's openness for accepting diverse solutions the Member States have.

It has been common practice to carry out scientific research on previously collected samples and data. Here, a biobank framework that focuses on the samples and associated data stands, at least on the surface, in contrast to the GDPR as reviewed in this article. The Recommendation on research on biological materials of human origin prescribes detailed, rather narrowly constructed, provisions for

[161] Recommendation on the protection of health-related data, *op. cit.*, para.6.

[162] Currently, the central instrument in the EU in that regard is Council Directive 2000/78/EC of 27 November 2000 establishing a general framework for equal treatment in employment and occupation OJ L 303, 2.12.2000, 16–22.

when research not covered under the initial consent is possible, and places the individual at the centre whenever possible. The GDPR, on the other hand, has placed consent as one of the research enablers. However, it is not necessarily the only one or a prioritised one. This also applies for other processing activities, such as data sharing in the course of scientific research. This essentially means that under the GDPR there are several avenues for obtaining controlled access to one's state of separateness and autonomous choice is only one of them. Considerable interventions in the state of separateness could be made, which are then balanced out by the right to information and other rights. That right, when data are not obtained from the data subject, could be muted on the condition that appropriate safeguards are in place.[163] It is precisely situations such as these where the discussion on what is understood as appropriate safeguards gains particular importance and more clarity is needed regarding what complementary measures to safeguard the state of separateness are possible under the GDPR,[164] and that can be used to mute such important rights as right to information, which has a direct link to a bottom-up oversight mechanism. This discussion on the state of separateness is of particular importance in the context where the initial data are obtained at a pre-birth stage but subsequently get protected, and the initial step of accessing these data has not been under the control of the data subject and it has occurred through someone else's state of separateness.

The right to privacy and data protection have historically been intertwined and enjoyed a complex relationship.[165] Even though the GDPR sets forth requirements that are relevant for the state of separateness and can be seen as the EU's approach to reconciling various interests at stake as far as informational privacy is concerned, it does not *expressis verbis* focus on the protection of privacy or the protection of the right to private life. In Recital 1 it emphasises data protection as a fundamental right in the EU legal order under Article 8 CFREU and sets out everyone's right to the protection of personal data as protected under Article 16 TFEU. Protection of the right to private life came into play when the GDPR affirmed that it "respects all fundamental rights and observes the freedoms and principles recognised in the Charter as enshrined in the Treaties", which includes private life.[166] However, the CJEU in its jurisprudence has appeared to be less keen and in fact cut the link between private life and data protection.[167] This may be teething problems that will eventually be overcome or may point to a more sophisticated interplay that remains to be illuminated and elucidated in the future.

One can question the implications of the data protection regime when such a regime prescribes controlled access to informational privacy. Given that the GDPR does not prescribe quantitative or contextual limitations to access informational privacy related to fulfilment of requirements set forth in Article 6(1) and 9(2), one could argue that there are good preconditions for the field to head in the direction of genetic transparency. Growing data collections and enhanced availability of genetic in-

[163] GDPR, *op. cit.*, Article 14(5)(b).

[164] For some insights *see* A.M. DUGUET, J. HERVEG, *Safeguards and Derogations Relating to Processing for Scientific Purposes: Article 89 Analysis for Biobank Research*, in S. SLOKENBERGA, O. TZORTZATOU, J. REICHEL (eds) cit.

[165] J. KOKOTT, C. SOBOTTA, *The distinction between privacy and data protection in the jurisprudence of the CJEU and the ECtHR*, in *International Data Privacy Law*, 3/4, 2003, 222.

[166] GDPR, *op. cit.*, recital 4.

[167] *See* e.g. the case of 11 December 2019, C-708/18, TK v Asociaţia de Proprietari bloc M5A-ScaraA, ECLI:EU:C:2019:1064, paras.33, 47, and 52.

formation mandates continuous monitoring and assessment of the adequacy of the data protection mechanism to manage the state of separateness and calls for further debates on whether new approaches need to be looked for. Here, a particular role should be assigned to the EU on the arguable presumption that actions under the principle of conferral also bring responsibility for the adopted regulations in the field.

ISSN 2284-4503

Special issue

Collection and sharing of genomic and health data for research purposes: Going beyond data collection in traditional research settings

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ABSTRACT: In the recent years collection of health and genomic data for biomedical research purposes has been expanded beyond traditional research settings. In doing so, various online tools and platforms are being utilized to collect data from various sources including Electronic Health Records, mHealth applications, disease registries and patient generated databases. While there is relatively higher certainty regarding the legal grounds for processing health and genomic data in the traditional research setting, the questions remain about the applicable legal framework when collecting data from other sources. In addition, given the diverse nature of collected data, adhering to traditional care-research distinction to determine the applicable legal requirements is confronted with complexities. This is particularly the case when data collected in the care setting are being later used for research purposes. In this article, we discuss the challenges associated with governance of processing data collected outside research settings and underline the steps should be taken to ensure conformity of such data processing by the applicable data protection regulations.

KEYWORDS: Genomic data; biomedical research; mHealth; Real-World Data; GDPR

SUMMARY: 1. Introduction – 2. Data collection via mHealth applications and online platforms – 3. Data protection in the context of mHealth applications and online platforms – 4. Collection of Real-World Data (RWD) – 5. Data Protection framework for processing RWD – 5.1 Source of the initial data collection and a question of further processing of data – 5.2. Legal requirements for using RWD for research purposes – 6. Path forward.

1. Introduction

Genomic research requires access to a large scale of research and clinical data in order to improve the statistical power of the databases and assist finding similar cases. In the recent years, a need for access to large scale of data has led to increasing support for data sharing among researchers across the world. In particular, researchers who are funded by public funding are strongly recommended or in some instances mandated to share their genomic data through public genomic databases such as dbGaP (the database of Genotypes and Phenotypes) and

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EGA (European Genome-phenome Archive)[1]. This way, the data generated through public money can be put in optimal use and other researchers across the world can have the opportunity to run downstream analysis on the existing databases.

In addition, collection of health and genomic data outside the traditional research setting has received increasing attention. One of the important sources of such data is related to patients and individual-generated data which can be collected via various mHealth related applications and online platforms. This way, individuals could contribute health and genomic data either by uploading their genomic test results or submitting other health related information through filling questionnaires or self-measurement of various variables. This would also facilitate collection of so-called real-world data (RWD), namely data which are routinely collected outside a controlled research environment. As it has been stated in a recent report by the Duke Margolis Center for Health Policy: "Mobile health (mHealth) apps and wearables, particularly those that collect patient- and consumer-generated health data, can fill some of […] data gaps by providing real-world, more meaningful, high frequency, and/or longitudinal data." [2]

Real World Data have attracted an increasing attention in recent years due to the fact that evidence provided by traditional clinical research often fails to answer patients', physicians' and healthcare decision-makers' questions about real-world practices and outcomes. This is specially of interest in the context of clinical trials, as it has been expressed in a statement issued by Roche on Access to and Use of RWD: "While clinical trials focus on ensuring that valid causal conclusions can be drawn between intervention and effect, RWD are seen as a potentially rich and underutilized source to generate insight as to how approved diagnostics systems and medicines affect outcomes for patients under real world conditions."[3]

The collection and processing of health data for research purposes outside traditional research setting including using consumer health applications and other mobile devices, raises a number of privacy and data protection issues. Notably, privacy concerns are being intensified in the context of genetic data, as full anonymization of genetic data is not possible, owing to the nature of genetic data which contains unique identifiers about the individuals. In Europe, collection and use of personal data must be compliant with the EU General Data Protection Regulation (GDPR), and other applicable national regulations.

In the framework of the GDPR, which has been a main data protection legal instrument in the EU since its implementation in 2018, personal data is defined as any information relating to an identified or identifiable natural person. Therefore, processing data that have been irreversibly de-identified may fall outside the scope of the relevant personal data protection regulations. The GDPR does not specify the exact identifiers which need to be removed in order to render data non-identifiable, but notes that "to determine whether a natural person is identifiable, account should be taken of all the means reasonably likely to be used, such as singling out, either by the controller or by another

[1] M. Shabani M, B. Knoppers, P. Borry. *From the principles of genomic data sharing to the practices of data access committees*, in *EMBO Molecular Medicine*, 7, 2015, 507-509.

[2] Duke Margolic Center for Public Policy. Mobilizing mHealth Innovation for Real World Evidence Generation. Available online at: https://healthpolicy.duke.edu/sites/default/files/2020-03/duke-margolis_mhealth_action_plan.pdf.

[3] Roche, *Roche Position on Access to and Use of RWD*, 2019, available online at: https://bit.ly/32p5JWE.

ISSN 2284-4503

person to identify the natural person directly or indirectly."[4] Other regulations concerning health data, such as HIPAA (Health Insurance Portability and Accountability Act) in the USA, take a different approach, namely by listing specific identifiers that should be removed from datasets in order to de-identify them.[5]

The GDPR foresees, among other things, specific provisions regarding processing sensitive personal data, including health and genetic data, and provisions regarding processing data for scientific research purposes. In particular, it should be ensured that data processing is based on one of the recognized legal bases under the applicable data protection regulations. In addition, it should be ensured that adequate organizational and technical measures are adopted to mitigate the risks of re-identifiability of data and privacy breaches when processing individuals' data.[6]

In this manuscript, first, we provide an overview of emerging data collection approaches for biomedical research purposes outside traditional research setting, including collection of data via mHealth applications and online platforms, and collection of real-world data. In the following, we will discuss the associated legal concerns related to collection and processing of such data and analyze how the existing data protection framework would apply to the data collection and use outside traditional research setting, and what would be the future approaches regarding applicable legal framework for such data collection and data reuse.

2. Data collection via mHealth applications and online platforms

Smartphone applications for health are being increasingly used as a platform for collecting mass volumes of crowdsourced personal health data. Smartphones, for example, record and process numerous health measurements, including behavioral measures, clinical data, and health related symptoms. More than five billion people around the world own some kind of mobile device and in advanced economies, rates of smartphone ownership average nearly 80%.[7] A majority of smartphone owners have used their devices to access, record, or track data relevant to their health.[8] The popularization of personal health data monitoring, the increasing ubiquity of smartphone applications with health-related functions, and the rapidly improving technological capacities of mobile devices, have led to unprecedented opportunities for harnessing crowdsourced health and genomic data to expand biomedical knowledge. At the same time, the increasing popularity of consumer genetics products is permitting individual result processing while also facilitating the

[4] Recital 26, GDPR.

[5] M. SHABANI, L. MARELLI, *Re-identifiability of genomic data and the GDPR Assessing the re-identifiability of genomic data in light of the EU General Data Protection Regulation*, in *EMBO reports*, 20, 2019, e48316.

[6] Article 89 (2), GDPR.

[7] L. SILVER, *Smartphone Ownership Is Growing Rapidly Around the World, but Not Always Equally*, *Pew Research Center*, 2019, available online at: https://www.pewresearch.org/global/2019/02/05/smartphone-ownership-is-growing-rapidly-around-the-world-but-not-always-equally/.

[8] A. EDWARDS, *mHealth: Healthcare Mobile App Trends in 2019,* in *Ortho Live,* 2019, available online at: https://www.ortholive.com/blog/mhealth-healthcare-mobile-app-trends-in-2019.

sharing of such information with clinicians and health researchers,[9] potentially complementing and enhancing the utility of data collected on mobile device platforms.

The collection of genomic and health data via mHealth applications has been also considered as a citizen science activity, whereby the general public is directly involved in collection of data and conducting scientific research.[10] In the context of genomic research, this has been enabled by availability of Direct-to-Consumer genetic testing services which allow individuals to have access to their raw data and share that for research purpose. To name a few examples of such projects, Citsci.org, Open Humans.org, and the platform created by the for-profit company PatientsLikeMe are currently providing platform for citizen science-based data collection.[11] This will promote easier participation of the individuals in the research and facilitate efficient access to individual-generated data.

A study conducted by Talwar and colleagues have systematically reviewed the existing genetics and genomics apps in the market. They concluded that "while the majority of the apps served as references or resources (i.e., providing general genetics/genomics information and/or tutorials), some apps provided lifestyle recommendations to the general public, mostly based on the DTC genetic test results. Specifically, using integrated schemes and algorithms, those apps could provide an interpretation of genetic test results and then offer personalized recommendations regarding nutrition and physical activity." [12] This study identified eighty-eight genetics and genomics related apps, although it is not clear how many of these apps also collect and reuse users' data for research purposes. As developing and improving apps require access to a large scale of data, it is expected that the collected data from the users to be considered as a valuable resource for the app developers. This feature is a prominent aspect in the context of AI-driven technologies which use machine learning methods for finding a pattern in data.

In addition, other initiatives have been emerging which aim to collect personal genomic data from individuals in exchange for various financial incentives. The examples are Luna DNA, Nebula Genomics and ENCRYPGEN which invite individuals to upload their DNA data to be used for various research and clinical purposes by the interested parties. To make such data sharing by the individuals fair, they offer various incentives in exchange for data, including returning a free DNA report, DNA tokens or shares. [13] Although these platforms allegedly are utilizing innovative ways to collect personal genomic data, concerns remain regarding privacy and ownership, and compatibility of these approaches with the research ethics principles. We will discuss some of these concerns in the following part.

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[9] M CABELL *Jonas* et al*. Physician Experience with Direct-To-Consumer Genetic Testing in Kaiser Permanente*, in *Journal of Personalized Medicine,* 9:4, 47, 2019.

[10] E. HAFEN. *Personal Data Cooperatives – A New Data Governance Framework for Data Donations and Precision Health*, in: J. KRUTZINNA, L. FLORIDI (eds) *The Ethics of Medical Data Donation. Philosophical Studies Series*, in *Springer*, 137, Cham, 2019, https://doi.org/10.1007/978-3-030-04363-6_9

[11] M. MAJUMDER, A. MCGUIRE. *Data Sharing in the context of Health-related Citizen Science*, in *Journal of Law, Medicine, and Ethics*, 2020, https://doi.org/10.1177%2F1073110520917044.

[12] D. TALWAR. *Characteristics and quality of genetics and genomics mobile apps: a systematic review*, in *European Journal of Human Genetics,* 27 (6), 2019, 833-840.

[13] E. AHMED, M. SHABANI. *DNA Data Marketplace: An Analysis of the Ethical Concerns Regarding the Particopation of the Individuals,* in *Frontiers in Genetics, 2019*, https://doi.org/10.3389/fgene.2019.01107.

3. Data protection in the context of mHealth applications and online platforms

Protecting the rights of the individuals on their personal data including their right to privacy is a cornerstone in collection, storage, uses and sharing of health-related and genomic data. Collection of data from the individuals for research purposes may raise special data protection and privacy concerns. First, when collecting individual-generated data via various mHealth applications or other health data from individuals via online platforms, it is essential to ensure that individuals are fully aware of initial and secondary data use purposes. As we have mentioned in the previous section, it is likely that the data collected from the individuals via genomics and genetics related apps to be used for further development of the apps or improvement of the existing versions, or other relevant research purposes. According to the GDPR, the data subjects have a right to receive transparent information regarding how their data are being processed and for which purposes.[14] Often mhealth applications and online platforms develop privacy policies in which they include information regarding purposes of data processing and potential secondary uses, such as for research purposes. The previous investigations have shown that often individuals do not read long privacy policy documents or are not always fully informed about the subsequent changes to the privacy policy.[15] As a result, the adequacy of the privacy policies in ensuring transparency of the data processing has been questioned at times.

Second, the proposed models of DNA data marketplaces raise an array of concerns related to adequate legal and policy framework for protection of privacy and ownership rights of the individuals when exchanging their data for free test reports, DNA tokens, shares, and the like. Notably, offering compensation for donating data for research purposes has been traditionally considered questionable under research ethics principles, due to the concerns about undue influence on individuals and potentially rendering their consent invalid.[16] In addition, in view of a legal vacuum for data ownership in many jurisdictions, it remains to be seen how monetary value of data will be evaluated in a fair and legally valid manner and to be legally protected in case of future disputes. In terms of privacy, some of these emerging platforms are suggesting adopting new technologies such as blockchain to increase the security of the data processing in a decentralized manner. Blockchain is an emerging technology of a decentralized, digitized database medium and a public ledger of all transactions in the network.[17] Blockchain-based solutions have recently gained popularity in the context of genomic and health data sharing, with the promise of improving data access, patient empowerment, and improved interoperability. [18] That said, the implementation of blockchain technology in the context of genomic and health data sharing is still in its infancy and it remains to be seen how far this can address the data protection and data access governance concerns.

[14] Art 12 & 13, GDPR.

[15] N. STEINFELD. "*I agree to the terms and conditions": How do users read privacy policies online? An eye-tracking experiment. Computers in Human Behavior*, 55 (Part B), 2016, 992-1000.

[16] E. AHMED, M. SHABANI. D*NA Data Marketplace: An analysis of the ethical concerns regarding the participation of the individuals",* in *Frontiers in Genetics,* 2019, https://doi.org/10.3389/fgene.2019.01107

[17] H. OZERCAN, I., ILERI, E. AYDAY, C. ALKAN, *Realizing the potential of blockchain technologies in genomics*, in *Genome Res*, 28, 2018, 1255–1263, https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6120626/ .

[18] M. SHABANI. *Block-chain based platforms for genomic data sharing: a decentralised approach in response to the governance problems?,* in *JAMIA*, 2018, https://doi.org/10.1093/jamia/ocy149.

4. Collection of Real-World Data

Another type of health data which is of interest for biomedical research is so called real-world data (RWD) which are often collected outside controlled research setting. The rise of interest in RWD is driven by the increasing need for evidence in specific populations, such as comorbid or multi-treated people. In addition, RWD may represent the only source of information in some fields of special interest, e.g., rare diseases. RWD can also allow investigation of unanticipated, uncommon or long-term outcomes, and be integrated into the health technology assessments, in particular relative effectiveness assessments (REAs) and cost-effectiveness assessments (CEAs) of novel or existing drugs in clinical practice, thereby supporting Randomized Clinical Trials (RCT) evidence.[19]
Real world data can be collected from different sources. According to a definition provided by the US Food and Drug Administration (FDA), RWD may include "data related to patient health status and/or the delivery of health care routinely collected from: electronic health records (EHRs), claims and billing data, [data from] product and disease registries, patient-generated data including home-use settings, and data gathered from other sources that can inform on health status, such as mobile devices."[20]

5. Data Protection framework for processing RWD

As access to and use of RWD involves processing of sensitive health-related data collected from patients and research participants, it is crucial to investigate the lawful basis for processing RWD data and the relevant requirements when processing RWD for research purposes. Use of RWD may fall under the scope of relevant personal data protection regulations, if it is considered to be personal data. Notably, RWD may include both individual level patients' and research participants' data, but also aggregate data which would fall outside the scope of the GDPR. Processing individual level data that are considered as identifiable, consequently, needs to be on the basis of one of the lawful grounds recognized by the GDPR. In the Article 6 (1) of the GDPR the lawful grounds for processing personal data are listed, including obtaining consent, processing for compliance with a legal obligation, public interest, or legitimate interest. In addition, under Article 9 (2) of the GDPR, the special categories of data, including health data, can be processed on the basis of one of the recognized lawful bases, including obtaining explicit consent, among others. Furthermore, processing of special categories of data can be permitted if the processing is for scientific research purposes, and under the so-called research exemption provisions.
Processing RWD however may fall under various legal provisions, first depending on the source and a legal basis for the initial data collection and second, the specific purpose of using RWD. In the following, we will further elaborate these elements.

[19] A. MAKADY, et al. *Using Real-World Data in Health Technology Assessment (HTA) Practice: A Comparative Study of Five HTA Agencies,* in *Pharmacoeconomics*, 36, 3, 2018, 359-368, https://rdcu.be/ciOQb.
[20] FDA*, Real World Evidence*, 2020, available at: https://www.fda.gov/science-research/science-and-research-special-topics/real-world-evidence

5.1. Source of the initial data collection and a question of further processing of data

As it has been briefly explained earlier, RWD may refer to the data that are extracted from diverse sources, including electronic health records (EHRs), claims and billing data, data from product and disease registries, patient-generated data including home-use settings, and data gathered from other sources that can inform on health status, such as mobile devices. In this sense, use of RWD could be mainly based on the processing of the existing data, rather than new data collection. Therefore, the data controllers should clarify whether processing of RWD deviates from the original data collection purposes, thus considered as further processing of data, and whether such further processing is allowed by law. The GDPR allows further processing under a number of conditions:

"The processing of personal data for purposes other than those for which the personal data were initially collected should be allowed only where the processing is compatible with the purposes for which the personal data were initially collected. In such a case, no legal basis separate from that which allowed the collection of the personal data is required."[21]

One can argue that further processing of for example electronic health records or patient-generated data such as those gathered from mobile services, for clinical trials or HTA purposes may not immediately considered as compatible with purposes for which the personal data were initially collected. However, according to the GDPR, the further processing of data may also be regarded as compatible and lawful if the processing *"is necessary for the performance of a task carried out in the public interest or in the exercise of official authority vested in the controller, Union or Member State law".*

A question here is whether processing RWD can be considered for the purpose of the "exercise of official authority vested in the controller, Union or Member State Law". Our answer here is mainly dependent on how the nature of RDW use has been defined. RWD can be used to inform the assessment of the reliability, safety or relative effectiveness of a treatment. Since in principle this is part of the objectives of clinical trials, the questions arise whether processing of RWD is to comply with the legal obligations to which the sponsor and/or the investigator is subject to under the relevant clinical trials regulations. Looking at the EU Clinical Trials Regulations 2014 and a relevant opinion issued by the European Data Protection Board, it appears that not all data processing in the framework of clinical trials are considered to be for the purposes of compliance with legal obligations. In fact, besides the processing that is strictly necessary for safety reporting or archiving obligation of clinical data, the rest of processing is more likely to be considered as a research activity. [22]

In case processing of RWD is considered to be for scientific research purposes, then the processing should be in compliance with the relevant requirements set by a number of regulations. While some of these provisions are embedded in the applicable data protection regulations, others are set by regulations concerning human subjects research and clinical research.

[21] Recital 50, GDPR.

[22] European Data Protection Board, Opinion 3/2019 concerning the Questions and Answers on the interplay between the Clinical Trials Regulation (CTR). Available online at: https://edpb.europa.eu/our-work-tools/our-documents/avis-art-70/opinion-32019-concerning-questions-and-answers-interplay_en

5.2. Legal requirements for using RWD for research purposes

To begin with, it is important to note that processing data for scientific research purposes is being recognized as a lawful basis for processing of special categories of data under Article 9(2), and a compatible purpose for further processing of data under Recital 50 of the GDPR. Notably, the recital 50 appears to assimilate purpose specification and lawfulness in the case of reuse for the purposes of scientific research. To further elucidate this recital, the European Data Protection Supervisor (EDPS) has stated that: "We [EDPS] would therefore argue that, in order to ensure respect for the rights of the data subject, the compatibility test under Article 6(4) should still be considered prior to the reuse of data for the purposes of scientific research, particularly where the data was originally collected for very different purposes or outside the area of scientific research. Indeed, according to one analysis from a medical research perspective, applying this test should be straightforward." [23]

Furthermore, processing data for scientific research purposes should be in accordance with Article 89 of the GDPR based on Union or Member States laws, and subject to adopting adequate technical and organizational measures to safeguard the rights of the data subjects. To date, diverse approaches are being adopted in implementation of Article 89 across the Member States, in particular in terms of safeguards required, when processing data for research purposes. Notably, further requirements may still apply due to the fact that collection and use of data for biomedical research and in particular clinical research concerns other applicable regulations regarding human subjects' research.

Finally, it might be difficult to draw a clear distinction between research and care when processing RWD data. In fact, the use of RWD may not be only necessary for generating evidence to benefit research purposes, but also improve health care decisions, or for the purposes of so-called learning healthcare systems. As Budrionis and Bellika put it, the learning healthcare system, among other things, focuses on "exploring the potential of data collected in daily clinical practice as a source of up-to-date minimally biased population-specific knowledge, which could be implemented into clinical practice in a more agile manner than randomized controlled trials."[24]

Taking such uses into considerations, it will be difficult to clearly separate research from clinical practice when using RWD. Notably, this poses extra complexities regarding feasibility of drawing a clear line between using data for research, care, quality assurance and proving safety and efficiency of medical products purposes when using RWD. In addition, the growing interest in linking various existing health-related databases, from electronic health records to data collected through wearables and apps, further challenges holding to traditional distinction between various purposes in order to define the legal requirements for processing data.

Furthermore, this has implications for determining the rights and responsibilities of the involved parties. This is particularly pertinent in some respects, such as the requirements for consent or return of secondary findings, where the applicable legal frameworks differ based on the nature of the respected activity.

[23] European Data Protection Supervisor- Preliminary Opinion on Data Protection and Scientific Research- 01/2020 & EDPB- Opinion No 3/2019 .

[24] A. BUDRIONIS, J. BELLIKA. *The Learning HealthCare System. Where are we now? A systematic Review,* in *Journal of Biomedical Informatics,* 64, 2016, 87-92.

ISSN 2284-4503

6. Path forward

Biomedical research is significantly benefiting from processing health and genomic data which are collected outside traditional research setting. Use of various mHealth application and online platforms, combined with access to other data sources such as EHR and patient registries has expanded biomedical researchers' access to a wide range of patient generated data and enables secondary uses of existing non-research databases. It is expected that this trend to be continued in the future and to be further facilitated by accessibility of various mHealth applications.

Processing of personal data for biomedical research purposes needs to be in compliance with the applicable data protection regulations. As we have shown above, processing personal data collected outside traditional research settings is associated with complexities regarding the nature of data processing and the relevant legal grounds and requirements for processing data. As we have seen in the case of data collection via mHealth applications and online platforms, the individuals may not be fully aware of secondary uses of data for research purposes. In view of the shortcomings of current privacy policies in adequately informing individuals about potential secondary uses of data, it is crucial to utilize innovative approaches to enhance transparency of the data processing for research purposes.

Further involving individuals in sharing genomic and health data, by enabling them to share their data directly with the interested parties seems advantageous, as this allows the individuals to have say in the way their data have been further processed for research purposes. However, we should note that "individual control" on health data or data ownership rhetoric in the context of health data are associated with significant limitations. In many jurisdictions, there is no legal framework for health data ownership for the individuals. Furthermore, individuals may not be adequately informed about the implications of exchanging their health data for monetary and non-monetary incentives such as tokens, shares or free sequencing. For instance, they may not be able to fully withdraw their consent for sharing data once they received free sequencing or the like in exchange for sharing data.

Furthermore, collection of data from various sources, may render it difficult to maintain to the traditional distinction of care-research when determining the applicable legal framework for data processing and clarifying the roles and responsibilities of the involved parties. As we have shown in this paper, for instance, the RWD can be collected both from existing clinical or patient-generated databases, to be used for various purposes including research purposes or informing the clinical decision-making. In that sense, such data processing may not strictly fit into one specific category. Therefore, rather than holding to traditional care-research distinction to determine the rights and responsibilities of the involved parties, it is crucial to identify the intricacies of data processing arising from secondary uses of data and adopt adequate safeguards in response to the associated risks. Tools and mechanisms such as Data Protection Impact Assessment (DPIA) which has been foreseen by the GDPR can be utilized in such assessment of the risks.

The next logical step would be adopting organizational and technical safeguards in the view of the identified risks. In this regard, taking advantage of emerging technological advancements in the area of data sharing and access such as distributed networks, which reduce a need for actual transfer of data is highly recommended. Furthermore, risks of re-identifiability of the individuals should be fully assessed and the adequate technical and organizational safeguards to be used to protect the data

subjects. Notably, the growing interest in connecting various research and clinical databases may pose new types of risks related to privacy and data protection.

Last but not least, when there is a question of further processing of data, which can lead to access to data by third parties, such as biotech or pharma companies, it is crucial to enhance transparency of data processing. To this end, the adequate information regarding the data processing should be communicated to the data subjects. Enhancing transparency when using patients and individual's data for research purposes would lead to higher trust on researchers and healthcare institutions.

ISSN 2284-4503

Special issue

Ethico-legal analysis of international sample and data sharing for genomic research during COVID-19: A South African perspective

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ABSTRACT: COVID-19 is a global pandemic that requires a global response, with a clear need for African involvement. Meaningful participation in global initiatives supports local knowledge-based opportunities for African researchers, builds local capacity and brings research in-house. However, the historical exploitation of vulnerable population groups within South Africa (SA) have given rise to legitimate ethical concerns including mistrust by communities when international transfers of samples and data are contemplated. Historical, cultural and ethical considerations have informed the development of regulations that apply to genomic research in many African jurisdictions. On 1 July 2020, SA's Protection of Personal Information Act No 4 of 2013 (POPIA) came into force, during an exponential rise of COVID-19 cases. Amid this evolving regulatory landscape, it is unclear what impact the South African regulatory framework will have on international sample and data sharing.

KEYWORDS: South Africa; international sample and data sharing; genomic research; COVID-19; broad consent; trust

SUMMARY: 1. Introduction – 2. The importance of South Africa's participation in pandemic research – 3. "Trust" in genomic data sharing – 4. Regulation of genomic research in South Africa – 5. Consent for genomic research in South Africa – 6. Broad consent under POPIA – 7. The use of broad consent for COVID-19 genomic research – 8. Legal status of international sample and data sharing – 9. Recommendations and Conclusion.

1. Introduction

Genomic research involves the use of biological samples and the generation of large data sets. The ease with which samples and data can be collected, used and re-used has brought about a shift in practice towards "open science" and a push towards the open sharing of biological samples, data and research results.[1] Open science can result in more reproducible science, encourages the optimal use of resources and can promote innovation using

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[1] M. WALPORT, P. BREST, *Sharing research data to improve public health*, in *Lancet*, 377, 2011, 538-539. See also : C. ALLEN, D.M.A. MEHLER, *Open science challenges, benefits and tips in early career and beyond*, in *PLOS Biology* 17, 12, 2019, 1-14.

existing data sets.[2] Despite the reported benefits of open science, there are tensions and challenges associated with sample and data sharing. In South Africa (SA), there are legal, ethical and historical factors which impact this type of sharing.

In SA, concerns regarding the use and international sharing of samples and data are perpetuated by the country's colonial past coupled with apartheid and power asymmetries in international collaborative research.[3] As occurred in many lower to middle-income countries (LMICs), in SA, local research facilities and researchers have been viewed as collectors of specimens only. Samples were then sent to high income countries (HICs) for use in research, with limited capacity development at the site of origin or local oversight of the sample at the research facility.[4] Attitudes towards the use of samples and data in these regions are influenced by previous experiences, but such practices are not consigned to the past. In 2019 a scandal emerged involving the Wellcome Sanger Institute who were accused of commercializing a gene chip without agreement from partner institutes or consent from hundreds of Africans who donated their DNA which was used to develop the chip.[5] This came not long after the controversies surrounding a phase I/II clinical trial of a candidate Ebola virus vaccine in sub-Saharan Africa was exposed, that involved a complete disregard of local regulatory procedures and secrecy approaches; practices that would be inconceivable in HICs.[6] Incidents such as these result in enormous setbacks to the trust relationship that is so necessary between researchers and participants, and researchers and sponsors.[7]

In addition to such power asymmetries, SA researchers are grappling with ethical issues that relate to sample and data collection, storage, use and sharing. Issues of privacy, confidentiality, autonomy, and the feedback of findings have and are currently being debated and explored in the context of genomic research in SA.[8] In addition, some African communities attach a deep cultural significance to their blood and human materials that must be respected.[9] These historical, cultural and ethical considerations have informed the development of regulations that apply to genomic research in

[2] M. WALPORT, P. BREST, *Sharing research data to improve public health*, cit., 538-539. See also: C. ALLEN, D.M.A. MEHLER, *Open science challenges, benefits and tips in early career and beyond*, cit., 1-14.

[3] K. MOODLEY, S. SINGH, *It's all about trust: reflections of researchers on the complexity and controversy surrounding biobanking in South Africa*, in *BMC Medical Ethics* 17:57, 2016, 1-9.

[4] B-J. HARDY, B. SEGUIN, R. RAMESAR, P.A. SINGER, A.S. DAAR, *South Africa: From Species Cradle to Genomic Applications*, in *Nature Reviews Genetics*, S19, S.20 ,2008.

[5] https://www.sciencemag.org/news/2019/10/major-uk-genetics-lab-accused-misusing-african-dna (last visited 29/01/2021).

[6] G.B. TANGWA, K. BROWNE, D. SCHROEDER, *Ebola Vaccine Trials, Chapter 6,* in D SHROEDER et al. (eds), *Ethics Dumping: Case studies from North-South Collaborations*, Switzerland, 2018, 49-60.

[7] P. TINDANA, S. MOLYNEUX, S. BULL, M. PARKER, *"It is an entrustment": Broad consent for genomic research and biobanks in sub-Saharan Africa*, in *Developing World Bioethics,* 19, 9, 2019.

[8] Academy of Science of South Africa Consensus Study, Human Genetics and Genomics in South Africa: Ethical, Legal and Social¸ Implications, 2018. Available at: https://bit.ly/3sH15hH (last visited 29/01/2021).

[9] K. MOODLEY, N. SIBANDA, K. FEBRUARY, T. ROSSOUW, *"It's my blood": ethical complexities in the use, storage and export of biological samples: perspectives from South African research participants*, in *BMC Medical Ethics,* 15, 4, 2017. See also: K. MOODLEY, S. SINGH, *It's all about trust: reflections of researchers on the complexity and controversy surrounding biobanking in South Africa*, cit., 1-9.

ISSN 2284-4503

many African jurisdictions. Regulations are at times precautionary and restrictive in nature,[10] very much focused on the samples themselves and often silent on the use and sharing of data.[11] This is changing with the emergence of general data protection regulations in many jurisdictions across the continent, and although not specific to the research sector, impact the use and sharing of data for genomic research.[12]

In SA, genomic research is regulated through the National Health Act No 61 of 2003, (NHA) its Chapter 8 Regulations, the SA Material Transfer Agreement (MTA) template and the 2015 Department of Health Ethics in Health Research guidelines (DoH ethics guidelines). The Protection of Personal Information Act No 4 of 2013 (POPIA) finally came into force on 1 July 2020 during an exponential rise of COVID-19 cases in SA and researchers have until 1 July 2021 to ensure that their data practices comply with the law. Like other data protection regulations such as the European Union's General Data Protection Regulation (GDPR), POPIA provides a high-level principle-based approach to the use of personal information,[13] that includes genomic data. It introduces strict requirements that must be met prior to the use of personal information and the transfer of data outside of SA.

The regulation of genomic research is thus changing with the coming into force of POPIA. This is a change that is occurring at a time of a global pandemic where there is a clear public interest in the rapid access to and sharing of personal information, both within SA and across its borders, to respond to COVID-19. Such sharing is important to better understand disease pathogenesis, for the development of treatment options, for vaccine development and to provide for more effective and humane care. An improved understanding of the pathogenesis of the disease may assist in identifying individuals who are at risk of contracting COVID-19 or of developing more severe diseases.[14] Several local and international consortia have been launched to better understand the genetic determinants of susceptibility to SARS-CoV-2 and COVID-19 disease severity from the perspective of both the host and the virus. From the outset of this pandemic, results and data have been shared.[15] Open science has become the norm,[16] and many journals and funders have

[10] C. STAUNTON, J. DE VRIES, *The Governance of Genomic Biobank Research in Africa: Reframing the Regulatory Tilt,* in *Journal of Law and Biosciences*, 2020, 1-20.

[11] J. DE VRIES, S.N. MUNUNG, A. MATIMBA, S. MCCURDY et al., *Regulation of Genomic and Biobanking Research in Africa: A Content Analysis of Ethics Guidelines, Policies and Procedures from 22 African Countries*, in *BMC medical ethics,* 8, 18, 2017, 1-9.

[12] C. STAUNTON, R. ADAMS, D. ANDERSON, T. CROXTON et al., *Protection of Personal Information Act 2013 and data protection for health research in South Africa*, in *International Data Privacy Law*, 10, 2, 2020, 160–179.

[13] "Personal data" is the term used in the GDPR. "Personal information" is the term used in POPIA. Personal information will be the term used throughout this paper.

[14] The COVID-19 Host Genetics Initiative*, The COVID-19 Host Genetics Initiative, a Global Initiative to Elucidate the Role of Host Genetic Factors in Susceptibility and Severity of the SARS-CoV-2 Virus Pandemic,* in *European Journal of Human Genetics*, 28, 6, 2020, 715-718.

[15] The COVID-19 Host Genetics Initiative, *The COVID-19 Host Genetics Initiative, a Global Initiative to Elucidate the Role of Host Genetic Factors in Susceptibility and Severity of the SARS-CoV-2 Virus Pandemic*, cit., 715-718. See also: Y. HOU, J. ZHAO, W. MARTIN, A. KALLIANPUR et al., *New insights into genetic susceptibility of COVID-19: an ACE2 and TMPRSS2 polymorphism analysis*, in *BMC Med*, 18, 216, 2020, 1-8. See also: *The Public Health Alliance for Genomic Epidemiology* at https://pha4ge.org/ (last visited 29/01/2021). See also: E.J. GRIFFITHS, R.E. TIMME,

committed themselves to sharing COVID-19 data and results.[17] This is a global pandemic that requires a global response and it is essential that SA participates in such research. However, in the midst of its evolving regulatory landscape, it is unclear what impact the South African regulatory framework will have on data sharing during a public health emergency (PHE).

This paper considers the effect of SA's regulatory framework on data sharing for COVID-19 genomic research. It begins by providing an overview of the importance of SA's participation in pandemic research and then considers trust as an ethical concept in genomic data sharing. It then explores consent implications on genomic data sharing, with a focussed discussion on the permissibility of broad consent during COVID-19. The paper further addresses whether the SA regulatory framework allows for the international sharing of data for COVID-19 genomic research and finally, it provides recommendations to the development of a Code of Conduct for Research to ensure more equitable and ethical research.

2. The importance of South Africa's participation in pandemic research

COVID-19 presents SA with an opportunity to contribute towards and participate in a priority global project. As evidenced by the inefficacy of AstraZeneca's vaccine against the South African variant,[18] if vaccine development is only focused on SARS-CoV-2 and its variants found in the global north, then there is a possibility that vaccines may not be effective in Africa. Dosages and frequency as well as transport and storage must be tailored to suit African populations across the differing regions in order to ensure efficacy.[19] Scientific validity in genetic studies depends in part on the amount of data that can be analysed and shared as a collective effort to ensure statistical significance.[20] This needs to be combined with an efficient and rapid analysis, particularly if this is to be effective during a pandemic. This sharing must continue as the virus evolves and new variants continue to be reported across the world. Indeed, it was through the sharing of samples and data that the SA variant that emerged at the end of 2020, was detected.[21]

A.J. PAGE, N-F. ALIKHAN et al., *The PHA4GE SARS-CoV-2 Contextual Data Specification for Open Genomic Epidemiology*, 2020, (https://www.preprints.org/manuscript/202008.0220/v1).

[16] J. HOMOLAK, I. KODVANJ, D. VIRAG, *Preliminary analysis of COVID-19 academic information patterns: a call for open science in the times of closed borders*, in *Scientometrics*, 124, 2020, 2687–2701.

[17] Sharing research data and findings relevant to the novel coronavirus (COVID-19) outbreak, Available at: https://wellcome.org/coronavirus-covid-19/open-data (last visited 29/01/2021).

[18] J. COHEN, *South Africa suspends use of AstraZeneca's Covid-19 vaccine after it fails to clearly stop virus variant,* in *Science*, 2021, available at: https://bit.ly/32F17vM (last visited 29/03/2021).

[19] K. CHIBALE, *Africa should be at the forefront of Covid Vaccine trials – and should be providing scientific leadership*, in *The Daily Maverick*, available at: https://www.dailymaverick.co.za/opinionista/2020-07-30-africa-should-be-at-the-forefront-of-covid-19-vaccine-trials-and-should-be-providing-scientific-leadership/ (last visited 29/01/2021).

[20] C.L. BORGMAN, *The conundrum of sharing research data*, in *Journal of the American Society for Information Science and Technology*, 63, 6, 2012,1059–1078.

[21] H. TEGALLY, E. WILKINSON, M. GIOVANETTI, A. IRANZADEH et al., *Emergence and rapid spread of a new severe acute respiratory syndrome-related coronavirus 2(SARS-CoV-2) lineage with multiple spike mutations in South Africa*, in *BMJ Yale*. Pre-print available at: https://www.medrxiv.org/content/10.1101/2020.12.21.20248640v1.full (last visited 20/01/2021). See also: C.K. WIBMER, F. AYRES, V.T. HERMANUS, M. MADZIVHANDILA et al., *SARS-CoV-*

Outside of the scientific need for involvement, meaningful participation supports local knowledge-based opportunities for African researchers, builds local capacity and brings research in-house. This, in turn, assists with preparing local infrastructure for future pandemics. Currently however, not all African researchers and research institutions have the required tools and infrastructure necessary to rapidly process data during a pandemic. The Africa Centres for Disease Control and Prevention (Africa CDC) has committed to capacity building and has collaborated with the European & Developing Countries Clinical Trials Partnership (EDCTP) and more recently, the Foundation for Innovative New Diagnostics (FIND). The EDCTP collaboration will contribute towards the development of epidemiologists and biostatisticians who can collectively conduct surveillance, public health research and support timely responses to disease outbreaks in Africa in the future.[22] The collaboration with FIND aims to build technical capacity in readiness for the introduction of new, high-quality antigen rapid diagnostic tests (RDTs) for COVID-19.[23] Monitoring interventions during the early stages of a pandemic is critical to prioritising future control efforts.[24] These collaborations, infrastructural development and personnel development not only ensures a local response to a global pandemic but will go towards ensuring that African jurisdictions have the infrastructure in place to respond to future pandemics. However, an effective response in a pandemic requires meaningful collaboration between all stakeholders including the scientific community, affected population groups and policy makers.

3. "Trust" in genomic data sharing

Trust is central to the legitimacy of health research systems and it forms the basis to which the social contract between researchers and participants is honoured.[25] It can ensure acceptance of and compliance with preventive or curative interventions, that can include the uptake of vaccines as well as changes in individual behaviours to reduce risk.[26] In the SA context, mistrust appears to be linked to the country's broader socio-political context including racial discrimination under apartheid and the continued marginalisation of vulnerable groups.[27] In addition, questions around what constitutes

2 501Y.V2 escapes neutralization by South African Covid-19 donor plasma. Pre-print available at: SARS-CoV-2 501Y.V2 escapes neutralization by South African COVID-19 donor plasma (biorxiv.org).

[22] EDCTP and Africa CDC collaborate to develop capacity for outbreak and epidemic response in sub- Saharan Africa. Available at: https://africacdc.org/news-item/edctp-and-africa-cdc-collaborate-to-develop-capacity-for-outbreak-and-epidemic-response-in-sub-saharan-africa/ (last visited 29/01/2021).

[23] EDCTP and Africa CDC collaborate to develop capacity for outbreak and epidemic response in sub-Saharan Africa. Available at: https://africacdc.org/news-item/africa-cdc-find-partner-to-build-capacity-for-covid-19-rapid-diagnostic-tests-in-africa/ (last visited 29/01/2021).

[24] A. CORI, C.A. DONNELLY, I. DORIGATTI, NM. FERGUSON et al., *Key data for outbreak evaluation: building on the Ebola experience*, in *Philosophical transactions of the Royal Society of London. Series B, Biological sciences*, 372, 1721, 2017, 1-14.

[25] A. KERASIDOU, *The role of trust in global health research collaborations*, in *Bioethics*, 33, 2019, 495– 501.

[26] P. VINICK, P.N. PHAM, K.K. BINDU, J.B. BEDFORD et al., *Institutional trust and misinformation in the response to the 2018-19 Ebola outbreak in North Kivu, DR Congo: a population based survey*, in *The Lancet*, 19, 5, 2019, 529-536.

[27] S. THABETHE, C. SLACK, G. LINDEGGER, A. WILKINSON et al., *"Why Don't You Go Into Suburbs? Why Are You Targeting Us?": Trust and Mistrust in HIV Vaccine Trials in South Africa*, in *Journal of Empirical Research on*

meaningful community engagement and challenges in its implementation do little to build confidence in genomic research.[28] For instance, community mistrust in researchers and government authorities negatively impacted public health interventions in the DRC's North Kivu during the Ebola outbreak.[29] It is therefore of utmost importance that research practices during a PHE foster trust. COVID-19 genomic research is playing out in a world where there is evidence globally that the general public is reluctant to donate data and mistrust the idea of sharing with multiple users (doctors, researchers, governments).[30] There is also little familiarity with or awareness of the concepts DNA, genetics and genomics,[31] suggesting that the research community not only needs to be trusted by the public, but that urgent steps must be taken to actively communicate the importance of genomic research, data donation, and subsequent sharing, with communities.

It is not just the participant in research relationships where trust is important, but trust is also necessary between researchers who share samples and data. Data sharing practices should be cognisant of global perspectives, including African perspectives and experiences. Involvement of African researchers and samples and data in a global response to COVID-19 should be met with reciprocal benefits that include equity of access to diagnostics, therapies and vaccines. However, data sharing in the context of health emergencies is a challenge[32] and this is in part fuelled by a lack of trust in data sharing relationships.

Indonesia's refusal to share its H_5N_1 samples with the World Health Organisation (WHO) without a legally binding agreement outlining benefit sharing arrangements and intellectual property rights, points to a lack of trust of the motives of HICs and global authorities when samples and data are transferred from LMICs.[33] The concerns prompting these stipulations by Indonesia included that samples freely provided to and subsequently used by HICs for vaccine and product development are

Human Research Ethics, 13, 5, 2018, 525-536. For the purposes of this paper, vulnerability is defined in accordance with the South African National Department of Health Ethics Guidelines, 2015 as follows: "the diminished ability to fully safeguard one's own interests in the context of a specific research project; may be caused by limited capacity or limited access to social goods like rights, opportunities and power." Section 3.2 on page 26 of the same Guideline further outlines that "advanced age, very young age, personal or environmental factors like extreme poverty and ordinarily poor access to health care may increase vulnerability."

[28] C. STAUNTON, P. TINDANA, M. HENDRICKS, K. MOODLEY, *Rules of engagement: perspectives on stakeholder engagement for genomic biobanking research in South Africa*, in *BMC MED Ethics*, 19, 13, 2018.

[29] *Build trust to combat Ebola*, in *Nature*, 2019. Available at: https://www.nature.com/articles/d41586-019-00892-6 (last visited 29/01/2021)

[30] A. MIDDLETON, R. MILNE, M.A. ALMARRI, A.V. WEST et al., *Global perceptions of genomic data sharing: What shapes the willingness to donate DNA and health Data?* In *The American Journal of Human Genetics,* 107, 2020, 743–752.

[31] A. MIDDLETON, R. MILNE, M.A. ALMARRI, A.V. WEST et al., *Global perceptions of genomic data sharing: What shapes the willingness to donate DNA and health Data?,* cit., 743–752.

[32] S. ABRAMOWITZ, T. GILES-VERNICK, J. WEBB, J. TAPPAN et al., *Data sharing in public health emergencies: Anthropological and historical perspectives on data sharing during the 2014-2016 Ebola epidemic and the 2016 yellow fever epidemic, 2018 final report.* Available at: https://www.glopid-r.org/wp-content/uploads/2019/07/data-sharing-in-public-health-emergencies-yellow-fever-and-ebola.pdf (last visited 29/01/20201).

[33] X. ZHANG, K. MATSUI, B. KROHMAL, A.A. ZEID et al., *Attitudes towards transfers of human tissue samples across borders: An international survey of researchers and policy makers in five countries*, in *BMC Med Ethics*, 11, 16, 2010.

ultimately sold back to LMICs at unaffordable prices.[34] These issues have emerged once again during this pandemic. The absence of appropriate benefit sharing arrangements with LMICs, the early shortage of COVID-19 diagnostic material in Africa, and the scramble by many HICs for access to COVID-19 therapies and vaccines (so-called vaccine nationalism, in the case of the latter) to the disadvantage of LMICs, does little to assure equity of access to diagnostics, therapies and vaccines for COVID-19.[35] Some jurisdictions have bought more doses per person than required,[36] once again reminding us of the impact that skewed global power imbalances have on equitable access to a possible COVID-19 People's vaccine, where a vaccine is available free of charge with distribution based on need.

As a key principle, trust cannot exist as a standalone concept but rather needs to be incorporated into a governance system that is founded on accountability and transparency. This is particularly significant for pandemic research where fostering mutual trust remains a challenge.[37] One method of empowering and developing trust amongst LMICs through the research process is for them to be at the forefront of COVID-19 trials with ongoing involvement. The Access to COVID-19 Tools (ACT) Accelerator, or ACT-Accelerator, is a global collaboration formed to accelerate development, production and equitable access to COVID-19 tests, treatments, and vaccines.[38] COVAX, one of three pillars of ACT-Accelerator aims to ensure equitable access to COVID-19 vaccines.[39] SA's President Cyril Ramaphosa, was recently named the ACT-Accelerator Facilitation Council co-chair and as of December 2020, at least 184 countries have expressed willingness to participate in COVAX.[40] In addition, ACT-Accelerator recently published a framework for the governance of personal data that is used to respond to COVID-19, intending to complement existing national and international regulatory instruments to enable access to and use of data without compromising fundamental rights.[41] The principle of trust is emphasised and underpinned by a governance framework that includes respect for persons and communities; equity; transparency; accountability; privacy; engagement; and non-exploitation.[42]

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[34] S.E. Davies, *The duty to report disease outbreaks: of interest or value? Lessons from H5N1*,in *Contemporary Politics*, 17:4, 2011, 429-445.

[35] J.N. Nkengasong, N. Ndembi, A. Tshangela, T. Raji, *Covid-19 vaccines: how to ensure Africa has access,* 2020. Available at: https://www.nature.com/articles/d41586-020-02774-8 (last visited 29/01/2021). See also: *The Economist, Rich countries grab half of projected Covid-19 vaccine supply*, 2020. Available at: https://econ.st/3nreFot (last visited 29/01/2021).

[36] Oxfam International, *Small group of rich nations have bought up more than half the future supply of leading Covid-19 vaccine contenders*, 2020. Available at: https://www.oxfam.org/en/press-releases/small-group-rich-nations-have-bought-more-half-future-supply-leading-covid-19 (last visited 29/01/2021).

[37] N.S. Munung, P.C. Chi, A. Abayomi, M.O. Afolabi et al., *Perspectives of different stakeholders on data use and management in public health emergencies in sub-Saharan Africa: a meeting report*. Open letter awaiting peer review. Available at: https://wellcomeopenresearch.org/articles/6-11 (last visited 29/02/2021).

[38] https://www.who.int/initiatives/act-accelerator (last visited 29/01/2021).

[39] https://www.who.int/initiatives/act-accelerator/covax (last visited 29/01/2021).

[40] https://www.who.int/countries/ (last visited 29/01/2021).

[41] ACT-A Framework p2. available at: https://www.finddx.org/wp-content/uploads/2021/01/ACT-A-Dx-data-governance-framework_15.01.2021.pdf (last visited 29/01/2021).

[42] ACT-A Framework, cit., p3.

Trust is crucial for the international sharing of samples and data, however, a governance framework that enforces trust through transparency and accountability, coupled with meaningful community engagement will assist in the practical implementation of this ethical concept. While legitimate ethical tensions are fuelled by mistrust regarding the transfer of samples and data, there is no doubt as to the global necessity for rapid access to data during pandemics. For SA to meaningfully participate in COVID-19 research, its current regulatory framework needs to provide favourable conditions for the international sharing of samples and genomic data. Based on this rationale, we now explore the SA regulatory framework for the sharing of samples and data for genomic research.

4. Regulation of genomic research in South Africa

Genomic research is broadly permitted under the National Health Act 2003, its Chapter 8 Regulations[43] and the DoH ethics guidelines which has quasi-legal standing and is legally enforceable.[44] Together they act as SA's general ethico-legal health research framework. POPIA now forms part of this regulatory framework in the context of the use of personal information in research. Personal information is broadly defined as including information relating to an identifiable, living, natural person, implying that genomic data is also covered.[45] POPIA sets out rights and duties which are designed to safeguard personal information and applies to the particular activity of processing personal information. Thus, while human biological samples and data fall under the remit of the National Health Act 2003, its regulations and the DoH ethics guidelines, biological samples themselves fall outside of the remit of POPIA. Data that is derived from a biological sample would be considered "personal information" and thus falls under the remit of POPIA. However, the biological sample itself would appear to fall outside of this legislative scheme. Although POPIA only applies to information relating to an identifiable, living, natural person, the potentially identifiable nature of genetic information needs to be considered more carefully. There may be a fine line between the exact point at which a genetic sample, that is innately identifiable, becomes personal information as contemplated by the Act.

POPIA has strict processing requirements for personal information and specifically prohibits the processing of special personal information which includes health information and biometric data.[46] However, this general prohibition[47] does not apply if consent is provided; if the processing is for historical, statistical or research purposes to the extent that the purpose serves a public interest and the processing is necessary for the purpose; if the processing is for research and it appears impossible or involves a disproportionate effort to ask for consent; or, if the processing is authorised by the Information Regulator (an independent body empowered to monitor and enforce compliance)[48] with appropriate safeguards in place.[49] Section 32(5), specifies that personal

[43] Regulations relating to the use of human biological material. GN R 177 GG 35099 of March 2012.
[44] Sections 3.3 9 and 3.5.2.3.
[45] Section 1.
[46] Section 26(1).
[47] Although the Act includes other exemptions, the exemptions to this general prohibition that are mentioned are the most pertinent ones for COVID-19 genomic research.
[48] Section 39.

information regarding inherited characteristics may be processed for research, thus reinforcing the legitimacy of processing genomic data for research purposes.

The Act therefore provides that some of the strict processing requirements can be limited or exempted from, if the personal information is processed for research. In addition, the Information Regulator may grant exemptions to some of the conditions for the lawful processing of personal information if such processing is in the public interest or where there is a clear benefit to the data subject[50] or a third party.[51] "Research activity" is included as being in the public interest.[52] POPIA thus echoes the approach of many SA national reports that emphasise the importance of the use of data in research, in particular, genomic research.[53] While POPIA clearly recognises the importance of research, the impact of these exemptions and limitations on the processing of genomic data for research during COVID-19 is less clear.

5. Consent for genomic research in South Africa

Prior to the coming into force of POPIA, specific consent, broad consent, and tiered consent were all permitted consent models in SA.[54] Specific consent is consent to one study only. POPIA requires that personal information must be collected for a specific, explicitly defined and lawful purpose.[55] Thus, it clearly permits specific consent. Specific consent however, limits the use of samples and personal data to one study. Therefore, other consent models have been proposed and adopted for genomic research.

Broad consent is a consent model that permits the use of samples and/or data for current research, for storage and for possible research on future unspecified research purposes.[56] Samples and data can be re-used for research, subject to oversight and approval by a research ethics committee. This consent model has been subject to criticism on the basis that it is not truly informed consent and limits the ability of a participant to exercise their autonomous choice.[57] However, it is seen by the H3Africa Framework for Best Practice for Genomics Research and Biobanking in Africa, as ethically appropriate, provided it is supported by community engagement, appropriate governance and a

[49] Sections 27 (1)(a), (c), (d) and (2).

[50] For health research purposes, a data subject under POPIA is referred to as a "research participant". Thus, for purposes of this paper, data subject and research participant are used interchangeably.

[51] Sections 37(1)(a)&(b).

[52] Section 37(2)(e).

[53] Department of Science and Technology Annual Report 2013/2014. Available at: https://www.gov.za/sites/default/files/gcis_document/201409/dstannualreport201314.pdf. (last visited 29/01/2021). See also: Academy of Science of South Africa Consensus Study, *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications*, 2018.

[54] As expressly stated in the 2015 Department of Health Ethics guidelines, Principles, Processes and Structures, para 3.3.6.

[55] Section13.

[56] D. WENDLER, *Broad versus Blanket consent for research with human biological samples*, in *Hastings Cent Rep.* 43, 5, 2013, 3-4.

[57] M. SHEEHAN, *Can broad consent be informed consent?*, in *Public Health Ethics*, 4, 3, 2011, 226-235.

mechanism that supports accountability and equity in the use of resources.[58] In the context of COVID-19, the ACT-Accelerator Framework provides for the processing of personal data without informed consent if it is necessary "for reasons of public interest in the area of public health[59]"and where there are "suitable specific measures in place to adequately safeguard the rights and freedoms of data subjects".[60]

Tiered consent offers a participant a series of choices about the research, type of research, subsequent use of samples, and level of data and sample sharing that they agree to.[61] By providing a range of options, it is argued that participants are more easily able to exercise an autonomous choice. But, the use of tiered consent could become restrictive as researchers may not be able to specify the types of research to be performed years down the line. In addition, as science and technology advance, it may become unviable to continue with the same types of research as outlined at the time the initial tiered consent was taken. Furthermore, it is often an option that a tiered consent model includes an element of broad consent in that participants may opt to select their samples and data to be used for further general health research purposes. Thus, the use of a tiered consent model is often contingent on the acceptability of broad consent.

6. Broad consent under POPIA

SA's NHA provides a framework for a structured uniform health system, taking constitutional obligations and other laws into account. While a general standard of disclosure is provided for in the NHA, the DoH ethics guidelines established in accordance with section 72(6) of the NHA expressly permits the use of broad consent for health research. However, uncertainty arises as to the permissibility of broad consent under POPIA. According to the DoH ethics guidelines, the nature of the further usage under broad consent should be described as fully as possible and should stipulate that further prior ethics review of the new study is necessary. Permission may be sought to re-contact the person if intended future use is outside the scope of the current consent.[62]

POPIA requires specific consent, but section 15(1) provides for the further processing if this further processing (or secondary use) is compatible with the original purpose for which it was collected. POPIA also states that further processing for research purposes is permitted if: (1) processing is necessary to "prevent or mitigate a serious and imminent threat to" public health[63] (we call this, research on the grounds of public health); (2) processing is necessary to prevent or mitigate a serious

[58] *Ethics and Governance Framework for best practice in genomic research and biobanking in Africa*, 2017, Available at: https://h3africa.org/wp-content/uploads/2018/05/Final-Framework-for-African-genomics-and-biobanking_SC-.pdf (last visited 29/01/2021).

[59] ACT-A Framework, cit., para.4.3.2, p5.

[60] ACT-A Framework, cit., para.4.3.2, p5.

[61] V. NEMBAWARE, K. JOHNSTON, A.A. DIALLO, M.J. KOTZE et al., *A framework for tiered informed consent for health genomic research in Africa*, in *Nature Genetics*, 51, 2019, 1566–1571. See also: E.M. BUNNIK, A.C. JANSSENS, M.H SCHERMER, *A tiered-layered-staged model for informed consent in personal genome testing*, in *European Journal of Human Genetics,* 21, 6, 2013, 596-601.

[62] DoH ethics guidelines, cit., para 3.3.6, p43.

[63] Section15(3)(d)(i).

threat to "the life or health of the data subject or another individual";[64] or (3) if the personal information is to be used for research purposes and "will not be published in an identifiable form"[65] (we call this, the general research justification). The second ground (i.e. the life or health of the research participant) would appear to apply to the use of an individual's data and would likely apply in the health context, but would not be suitable as a ground for health research that requires the use of large quantities of data. Thus, it would appear that for genomic research in a pandemic, the further processing of personal information is permitted for research on two grounds: (1) research on the grounds of public health; and (2) the general research justification.

There is however, an ongoing debate regarding the legal permissibility of broad consent under POPIA. Thaldar *et al.* argue that the Act should be strictly interpreted, and that specific consent is a prerequisite for research on genomic information.[66] Staunton *et al.* on the other hand argue that a purposive interpretation of POPIA permits broad consent.[67] A purposive interpretation takes into account that the right to privacy can be subject to justifiable limitations. The preamble to POPIA itself states that "economic and social progress" may require the "removal of unnecessary impediments to the free flow of information including personal information". This also takes into consideration that POPIA provides for exemptions to some of the strict processing requirements if the personal information is to be used for research. This debate has real implications for COVID-19 genomic research.

7. The use of broad consent for COVID-19 genomic research

In a 2020 guidance note on the processing of personal information for the management and containment of COVID-19, the Information Regulator recognised the need to process personal information of research participants to curb the spread of the pandemic.[68] However, no further guidance or clarity was provided as to whether broad consent is permitted under POPIA. Should POPIA permit specific consent only, this would mean that already approved studies would have to ensure re-contact and re-consent from participants in order to process samples and data lawfully and that data could not be re-used for other COVID-19 related studies. This may be challenging and will certainly stifle pandemic research where the rapid access to samples and data is essential. A purposive interpretation of the Act is in line with the SA ethico-legal framework; such an interpretation recognises the real need for data-sharing in a public health emergency, and this interpretation is also in accordance with international norms and standards on this point.

[64] Section 15(3)(d)(ii).

[65] Section15(3)(e).

[66] D.W. THALDAR, B. TOWNSEND, *Genomic research and privacy: A response to Stanton et al*, in *South African Medical Journal*, 110, 3, 2020, 172-174.

[67] C. STAUNTON, R. ADAMS, M. BOTES, E.S. DOVE et al., *Safeguarding the future of genomic research in South Africa: Broad consent and the Protection of Personal Information Act 4 of 2013* in *South African Medical Journal*, 109, 7, 2019, 468-470.

[68] Guidance Note on the Processing of Personal Information in the Management and Containment of Covid-19 Pandemic in terms of the Protection of Personal Information Act 4 of 2013. Available at: https://www.justice.gov.za/inforeg/docs/InfoRegSA-GuidanceNote-PPI-Covid19-20200403.pdf (last visited 29/01/2021).

Owing to the uncertainty regarding the legal status of broad consent, researchers may opt to apply for an exemption from one of the processing requirements (in this case the requirement of purpose specification). The Information Regulator may grant an exemption to a processing requirement under section 37 if she is satisfied that the public interest outweighs the interference with the privacy of the research participant or a third party. The public interest is stated as including research. While this would provide clarity, guidance on applying for exemptions has not yet been gazetted, and such a process is likely to take time.

Following the above analysis, the use of personal information for COVID-19 research could thus be done on one of four grounds. First, specific consent is clearly permitted, but it prevents the further use of the personal information and the sharing of the personal information if the third party with whom the researcher is sharing the information is not known at the time of collection. Second, an exemption can be applied for under section 37, but owing to the uncertain procedure and uncertain timeframe, that is impractical in this public health emergency. Third, broad consent is permitted under the general research justification. Fourth, broad consent is permitted for COVID-19 research on public health grounds.

Clarity on the legal status of broad consent is essential and a POPIA Code of Conduct for Research must resolve this. To this end, the Academy of Science of South Africa (ASSAF) which represents SA in the international community of science academies,[69] has begun working towards the development of a Code of Conduct for Research to guide the application of POPIA to research.[70] It is hoped that the Code will be ready and approved by July 2021, in order to avoid the resulting legal impediments on necessary and ethically required and justified research. In the meantime, SA and the rest of the world are in the midst of the devastation caused by the COVID-19 pandemic, and genomic research on COVID-19 is essential.

We believe that when considering POPIA as a whole, broad consent for COVID-19 genomic research is permitted. It is now important to consider the two grounds under which broad consent is justified. Looking first at the general research ground that permits broad consent, it states that the personal information cannot be published in an identifiable form. Genomic data is innately identifiable even if it is de-identified, thus genomic data is likely to be considered identifiable under POPIA. However, POPIA does not define what is meant by "published" and what impact this has on how the personal data is made available. We submit, however, that this requirement not to publish personally identifiable personal information would prohibit making this personal information available via a publically accessible database. Thus, personal information may not be used for any research, collaboration, or be subject to a funding contract or journal requirement that requires the deposition of identifiable personal information in a publically accessible database. To enable researchers in SA to deposit personal information in a database, there must be some mechanism in place to ensure that the personal information is not published. A database that is controlled by an independent Data Access Committee (DAC) and provides access subject to the requirement that the personal information will never be published, would appear to be POPIA compliant. The second public health

[69] https://www.assaf.org.za/index.php/about-assaf/about-assaf (last visited 29/01/2021).

[70] ASSAF steering committee and drafting group for a Code of Conduct for Research under POPIA, formalised in December 2020.

ground has no similar stipulation, but owing to the need to safeguard the rights of the research participant, we argue that access to personal information for research on the grounds of public health must equally be subject to approval by a DAC. Such clarity should be given by the POPIA Code of Conduct for Research, but we consider that such an approach ensures an appropriate balance between safeguarding participant rights and enabling access to personal information for research.

8. Legal status of international sample and data sharing

Broad consent allows the further processing or secondary use of the samples and data, that includes the sharing of the samples and data, subject to REC approval. However, the regulatory framework as to the international sharing of these samples and data needs to now be considered. In 2012, a number of Regulations to Chapter 8 of the NHA were published. One of these regulations relates to the import and export of human tissue, blood, blood products, cultures cells, stem cells, embryos zygotes and gametes.[71] An export permit in relation to samples is required; however, the Regulations are silent as regards data. In July 2018, a national MTA template[72] was published by the national Department of Health and prior to POPIA coming into force, was the only ethico-legal document that regulated the transfer of samples and data where extensive networking is contemplated. However, while POPIA safeguards the processing of data only, the national MTA template provides for both samples and data. It further incorporates benefit sharing arrangements and the regulation of secondary use, allows broad consent with HREC oversight and indicates that custodianship should remain with the providing institute. The MTA template is a living document and although academic debate has unfolded since its publication,[73] it is currently the only national template available which aims to protect institutions, researchers and participants when human material is transferred out of SA.

Section 72 of POPIA outlines conditions for the transfer of personal information outside of SA and there are five possible grounds, of which three are only likely to be possible in the case of research. First, transfer can take place if the law in the jurisdiction of the recipient country provides an adequate level of protection that upholds principles that are substantially similar for the processing of personal information.[74] This can be in the form of a law, or binding corporate rules, or a binding agreement between the parties.[75] Second, the research participant consents to the transfer. Third, if the transfer is for the benefit of the participant and consent to the transfer is not reasonably practicable to obtain, recognizing that if it were reasonably practicable, then the participant would be likely to provide it.[76] As it is written, Section 72 of the Act appears to suggest that if the transfer

[71] GN R 182 in Government Gazette 35099 of 2 March 2012.

[72] National Health Act 61 of 2003. Material Transfer Agreement of Human Biological Materials. Government Gazette No. 41781: 719, 20 July 2018. https://www.gov.za/sites/default/files/41781_gon719.pdf (Last visited 29/01/20201).

[73] D.W. THALDAR, M. BOTES, A.G. NIENABER, *South Africa's new standard material transfer agreement : proposals for improvement and pointers for implementation*, in *BMC Medical Ethics*, 2020, 21, 85.

[74] Section 72(1)(a).

[75] Section 72(1).

[76] Section 72(1)(e).

satisfies one or more of the above set out grounds, then it is permissible. Thus, for example, if the transfer is subject to consent or any of the other grounds, there is no requirement that the recipient country has similar protections in place. However, under Section 57(1)(d), if special personal information, or the personal information of a minor is to be transferred to a country that does not provide an adequate level of protection for processing personal information contemplated under section 72, the prior authorisation of the Information Regulator is required. Section 57(3) states that this prior authorisation will not be needed if a Code of Conduct has come into force for a specific sector. Thus, the forthcoming Code of Conduct for Research may alter this somewhat. However, owing to the importance of the constitutional right to privacy and the sensitivity of the personal information, we would strongly urge that, at a minimum, transfer of sensitive personal information must take place subject to a MTA that requires the personal information to be protected in line with POPIA.

Turning now to the possible grounds for transfer for COVID-19 research. Consent is only practical if the research participant was informed who the third party is with whom the data will be shared and the risks associated with that sharing. Owing to the realities of the pandemic, this is unlikely to be known at the time of collection. In the context of the international sharing of data, the countries to whom research teams may want to share may not be known at the time of consent. Furthermore, as per section 11(2)(b), the research participant must be able to withdraw their consent at any time. If there are no mechanisms in place to respect this withdraw after transfer has taken place, then consent is not a ground that can be used.

The second ground is that the transfer benefits the research participant and they would be likely to provide consent if they could. Decisions on this would need to be made per research participant as the transfer is for the benefit of the individual research participant. Thus, it would need to be demonstrated that the transfer is for the benefit of each individual research participant. This is unlikely to be practical or possible where large data sets are required to be transferred. The Responsible Party would need some basis on which to make this judgment, otherwise risk being sued.

Thus, for COVID-19 genomic research, initial and onward transfers can likely only take place if there is a similar level of legislative protection in place (such as countries regulated by the European Union's General Data Protection Regulation)[77], or if the recipient in the third country agrees to be subject to a binding agreement or corporate rules which provide an adequate level of protection, for example, a Data Transfer Agreement (DTA) and a data management plan. Decisions on whether there is an adequate level of protection in the third country's legislative framework will need to be made by the Responsible Party. This assessment will be challenging as the Information Regulator has not issued any guidance regarding what levels of protection it considers as adequate. For now, researchers will likely rely upon the guidance of the European Data Protection Board (EDPB) in carrying out this

[77] D. HALLINAN, A. BERNIER, A. CAMBON-THOMSEN , F.P. CRAWLEY et al., *International transfers of health research data following Schrems II: A problem in need of a solution*. (September 7, 2020). Available at SSRN: https://ssrn.com/abstract=3688392 or http://dx.doi.org/10.2139/ssrn.3688392

assessment.[78] This assessment is going to require expertise that may not be budgeted for. Going forward, institutions and research teams will need to ensure that resources are dedicated to such assessments, but this does not resolve the need for such assessment now. This also leads us to another issue: speed. During a pandemic, rapid data sharing can be crucial, but such an assessment can take time. In addition, after this assessment, if the transfer includes the personal information of children, this requires prior authorisation by the Information Regulator, a process that also takes time. Processing this information may thus only occur after the Information Regulator completes its own investigation which could take up to 13 weeks to conduct. This could defeat the purpose of rapid access to and sharing of personal information of children during a pandemic.[79] This need for rapid data sharing in a public health emergency must be balanced with the need to protect the personal information of children. The forthcoming POPIA Code of Conduct for Research should provide guidance on how best to achieve this balance.

If after this assessment, a country is found to not have a similar level of protection through its laws, there must be either a binding corporate rule or binding agreement in place prior to transfer. Unlike the European Union, there are no standard contractual clauses[80] for transferring personal information out of SA, which have been a key legal mechanism for the transfer of personal data out of the European Economic Area (and more recently, the UK) for almost two decades. Thus, researchers will need to rely on DTAs. There is no standard DTA available in SA for researchers to use when transfers of personal information are contemplated. Currently, MTAs, an export permit, and an informed consent document are required by the office of the DoH when human biological materials are transferred outside SA. There is no uniform DTA template, which incorporates the safeguards that POPIA places on data transfer, available for researchers to populate. It may be argued that the current SA MTA template, which includes "data" into its definition of materials, could be used as a DTA; however, the provisions within the MTA template are more useful to the transfer of samples as the intricacies and technical aspects regarding data have not been incorporated into the document. The current MTA may be used as a guide, but researchers must ensure that they require an MTA in advance of sharing. The forthcoming POPIA Code of Conduct for Research must also provide guidance on the DTA and what is to be included. For now, it is clear that if a recipient in a third country does not have an appropriate level of protection in place, transfer for COVID-19 research must take place subject to a DTA, in addition to the other requirements stipulated by the DoH.

9. Recommendations and Conclusion

SA has a duty to participate in and contribute towards international collaborative research in public health emergencies, for the benefit of its diverse population groups. While legitimate ethical tensions

[78] European Data Protection Board Recommendations 01/2020 adopted on 10 November 2020. Available at: edpb_recommendations_202001_supplementarymeasurestransferstools_en.pdf (europa.eu) (last visited 08/04/2021).

[79] Sections 58(2) and (4).

[80] GA4GH GDPR Brief: *International "onward" transfers of genomic data under the EU Standard Contractual Clauses* (December 2020) Available at: https://www.ga4gh.org/news/ga4gh-gdpr-brief-international-onward-transfers-of-genomic-data-under-the-eu-standard-contractual-clauses/ (last visited 29/01/2021).

exist regarding mistrust towards the transfer of samples and data, participating meaningfully in global initiatives can assist with local capacity building and prepare local infrastructures for future pandemics, in the long term. SA's regulatory framework thus needs to be aligned accordingly to achieve this purpose. It is therefore imperative that the current uncertainties are remedied regarding what is or is not permissible for international data sharing during a pandemic. As the ASSAF works towards the development of a Code of Conduct for Research, it may consider providing guidance on how the global shift towards open science will be balanced with the safeguards and rights of research participants outlined within POPIA. In resolving some of the uncertainties in the application of POPIA to research generally, it must also consider research in a public health emergency and how to best balance the rights of the research participants with the need for rapid data sharing in a public health emergency.

As ASSAF continues with this work, we offer some tentative recommendations and points to consider, specifically for research during a public health emergency. First, under POPIA, genomic data can be processed if it is for research purposes. However, clarity is first required regarding the application of broad consent for health research purposes and specifically its permissibility for genomic research during a pandemic. We recommend that the Code should acknowledge that, while genetic data is innately identifiable, the appropriate consent model for health research should be aligned with SA's national ethics guidelines. POPIA must therefore be interpreted amongst pre-existing health regulatory frameworks and broad consent should be considered in addition to specific and tiered consent as provided for under the national ethics guidelines. Second, as POPIA does allow for broad consent for COVID-19 genomic research under the general research ground and public health ground, there should then rather be a shift in focus towards a suitable governance model that supports broad consent. Consideration should be given to DACs, their role and when a decision must be obtained from a DAC. We recommend that at a minimum, a DAC should oversee international transfers. Third, in the absence of guidance from the Information Regulator, clarity on how an assessment of a third country's data protection levels will take place and where the resources to fund such assessments will be sourced, must be contemplated, including expediting this process for research during a public health emergency. Fourth, it is essential that the Code should consider how the rapid transfer of sensitive personal information and the personal information of children should be managed during pandemics. Finally, we recommend making a DTA mandatory for international transfers. Guiding principles on standard provisions to be included within a DTA will also create a minimum standard for data transfers, which SA does not currently have. It is hoped that the forthcoming Code of Conduct for Research will provide much needed clarity not only for research during a public health emergency, but for research at a broader level, while fostering participation in open science as a benefit to SA.

ISSN 2284-4503

Special issue

The dynamic consent of the Cooperative Health Research in South Tyrol (CHRIS) study: broad aim within specific oversight and communication

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ABSTRACT: In biobanking and genomics research, data and samples are stored for long time and used in further studies, which may not be sufficiently specified or foreseen at the time of the initial consent. The dynamic consent of the CHRIS study integrates broad research aims, specific oversight and governance mechanisms, and continuous communication with participants, and allows nuanced choices to be changed over time. With this paper, we describe the CHRIS dynamic consent, and illustrate, by discussing data sharing and ongoing consent in the CHRIS study, how dynamic consent can actualize an informed consent model that is suitable for biobanking and genomic research.

KEYWORDS: Dynamic consent; biobanking research; oversight; communication

SUMMARY: 1. Which informed consent for biobanking and genomics research? – 2. Dynamic consent in practice: the case of the CHRIS study – 2.1. The CHRIS study – 2.2. Ten years of dynamic consent in the CHRIS study – 2.2.1. Dynamic informed consent for participation in the follow-up phase of the CHRIS study – 2.2.2. Information for an informed decision on participation in research – 3. Data sharing and ongoing consent in the CHRIS dynamic consent: broad consent within strong governance, oversight mechanisms, and ongoing information – 4. Conclusions.

1. Which informed consent for biobanking and genomics research?

Genomics and biobanking research have been generating considerable amounts of data and biological samples, which are invaluable dynamic resources for health research conducted through large size and long-term research projects. In biobanking, collected data and samples are stored and can be used for future studies, and can be shared in wider international and global research networks. This type of research poses ethical challenges: for example, given the long-term dimension of the research project and the rapid development of technology, the future uses of data and samples may not be defined nor foreseen at the time of initial consent, therefore

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providing an actual informed consent for the future uses of data and samples might result problematic. A debate on what kind of informed consent would better address the challenges posed by genomics research and biobanking is ongoing. Different consent procedures have been discussed, besides specific or broad consent, as possible approaches to meet the researchers' need for flexibility on the use of data and samples, and, at the same time, to protect participant autonomy and privacy, to protect against risks of harm, and to inform participants about the aims of the research and the risks and benefits of participation (e.g., solutions that envision broad consent coupled with governance and oversight mechanisms and provision of information, meta-consent, tiered consent, dynamic consent).[1]

Dynamic consent[2] has been proposed as a solution that allows both to meet the needs of the most recent biomedical research, and to comply with the ethical and legal requirements for participation in research. Dynamic consent is an interactive informed consent model. It is implemented through an online platform that allows a flowing communication between researchers and participants, and also a timely revision of the choices made regarding participation in research by participants. Dynamic consent is meant as a participant-centered[3] informed consent model which promotes participant autonomy, in particular in the case of long-term genomics projects, longitudinal studies, and biobanking research, where uses of data and samples may not be defined and explicitly foreseen at the time of recruitment. The flow of information will allow participants to be updated about further research developments and to make autonomous choices according to the changing circumstances in life and their values.

In the present paper, we describe the dynamic consent of the Cooperative Health Research in South Tyrol (CHRIS) study,[4] and we focus on two specific aspects of the CHRIS dynamic consent: data shar-

[1] E. Vayena, A. Blasimme, *Health research with big data: Time for systemic oversight,* in *The Journal of Law, Medicine & Ethics*, 46, 1, 2018, 119-129; C. Grady, L. Eckstein, B. Berkman, D. Brock, R. Cook-Deegan, S. M. Fullerton, H. Greely, M. G. Hansson, S. Hull, S. Kim, B. Lo, R. Pentz, L. Rodriguez, C. Weil, B. S. Wilfond, D. Wendler, *Broad consent for research with biological samples: Workshop conclusions*, in *The American Journal of Bioethics*, 15, 9, 2015, 34-42; D. Mascalzoni, A. Hicks, P. Pramstaller, M. Wjst, *Informed consent in the genomics era*, in *PLoS Medicine*, 5/9, 2008, 1302-1305. R.B. Mikkelsen, M. Gjerris, G. Waldemar, P. Sandøe, *Broad consent for biobanks is best - provided it is also deep*, in *BMC Medical Ethics*, 20, 71, 2019, 1-12; M. Sheehan, R. Thompson, J. Fistein, J. Davies, M. Dunn, M. Parker, J. Savulescu, K. Woods, *Authority and the future of consent in population-level biomedical research*, in *Public Health Ethics*, 12, 3, 2019, 225-236; T. Ploug, S. Holm, *Meta consent: a flexible and autonomous way of obtaining informed consent for secondary research*, in *BMJ*, 350, h2146, 2015, 1-4; H.J. Teare, M. Prictor, J. Kaye, *Reflections on dynamic consent in biomedical research: the story so far,* in *European Journal of Human Genetics*, Nov 28, 2020, 1-8.

[2] I. Budin-Ljosne, H.J. Teare, J. Kaye, S. Beck, H.B. Bentzen, L. Caenazzo, C. Collett, F. D'Abramo, H. Felzmann, T. Finlay, M. K. Javaid, E. Jones, V. Katić, A. Simpson, D. Mascalzoni. *Dynamic consent: a potential solution to some of the challenges of modern biomedical research*, in *BMC Medical Ethics*, 18, 4, 2017; J. Kaye, E. A. Whitley, D. Lund, M. Morrison, H. Teare, K. Melham, *Dynamic consent: a patient interface for twenty-first century research networks* in *European Journal of Human Genetics*, 23, 2, 2015, 141-146.

[3] J. Kaye, L. Curren, N. Anderson, K. Edwards, S. M. Fullerton, N. Kanellopoulou, D. Lund, D. G. MacArthur, D. Mascalzoni, J. Shepherd, P. L. Taylor, S. F. Terry, S. F. Winter, *From patients to partners: participant-centric initiatives in biomedical research*, in *Nature Reviews Genetics*, 13, 5, 2012, 371-376.

[4] The study protocol of the CHRIS study was described in C. Pattaro, M. Gögele, D. Mascalzoni, R. Melotti, C. Schwienbacher, A. De Grandi, L. Foco, Y. D'Elia, B. Linder, C. Fuchsberger, C. Minelli, C. Egger, L. S. Kofink, S. Zanigni, T. Schäfer, M. F. Facheris, S. V. Smárason, A. Rossini, A. A. Hicks, H. Weiss, P. P. Pramstaller, *The Cooperative*

ISSN 2284-4503

ing and ongoing consent. With these two cases, we illustrate how the dynamic consent of the CHRIS study actualizes a model in which broad and specific coexist, meaning that the broad aim of research is framed within strong governance and oversight mechanisms, and an ongoing specific communication, which allow participants to be informed and to make autonomous choices on participation in research through time.

2. Dynamic consent in practice: the case of the CHRIS study

The CHRIS dynamic consent was developed as a solution to meet the researchers' needs of conducting a long-term longitudinal biobank-based project, and the ethical and legal requirements for research in Italy, such as transparency and specific consent. In this section, we briefly describe the CHRIS study, and the dynamic consent as developed and implemented in the frame of the CHRIS study.

2.1. The CHRIS study

The CHRIS study is an ongoing longitudinal study, whose aim is to analyse the interplay among the genetic mechanisms, environmental factors, and human behaviour, which results in chronic conditions associated with human ageing in the general population of South Tyrol, Italy. The focus of the study is twofold: besides aiming to understand the etiological role of genetic and environmental risk factors on the onset and course of cardiovascular, neurological, metabolic, and oncologic diseases, the CHRIS study intends to promote awareness on the themes of health promotion and disease prevention.[5]

The CHRIS study started in 2011. The cohort was recruited in the middle and upper Val Venosta in South Tyrol, by inviting all the residents registered in the electoral list of 13 municipalities of the valley. The recruitment phase proceeded until December 2018, while the first follow-up phase begun in October 2019.[6] CHRIS researchers have been collecting biological samples (e.g., blood, urine) and data (anthropometric data and data on family history, lifestyle, and health, obtained through clinical

Health Research in South Tyrol (CHRIS) study: rationale, objectives, and preliminary results, in *Journal of Translational Medicine,* 13, 2015, 1-16. Further information on the CHRIS study can be found in the Eurac website (http://www.eurac.edu/en/research/health/biomed/projects/Pages/CHRIS.aspx last visited 16/01/2021) and in the dedicated CHRIS website (https://it.chris.eurac.edu/ last visited 16/01/2021).

[5] C. Pattaro, M. Gögele, D. Mascalzoni, R. Melotti, C. Schwienbacher, A. De Grandi, L. Foco, Y. D'Elia, B. Linder, C. Fuchsberger, C. Minelli, C. Egger, L.S. Kofink, S. Zanigni, T. Schäfer, M. F. Facheris, S. V. Smárason, A. Rossini, A. A. Hicks, H. Weiss, P.P. Pramstaller, *The Cooperative Health Research in South Tyrol (CHRIS) study: rationale, objectives, and preliminary results,* cit., 1-2.

[6] Since March 2020, when the COVID-19 emergency hit Italy, the CHRIS follow-up has been suspended. However, CHRIS researchers, in collaboration with Azienda Sanitaria dell'Alto Adige, developed and implemented the CHRIS-Covid-19 study, which started in July 2020. The study aims to understand the determinants of the infection and of viral transmission, and to study the immunity development and the long-term effects on health of SARS-CoV-2 infection (https://it.chris.eurac.edu/_startpage/chris-covid-19/, last visited 07/01/2021). The CHRIS follow-up is envisioned to re-start later this year when conditions for safety will allow.

examinations and questionnaires) from a closed cohort of more than 13000 adult participants.[7] All the data and samples are safely stored in the CHRIS biobank.

2.2. Ten years of dynamic consent in the CHRIS study

The dynamic consent of the CHRIS study can be defined as broad, for the broad description of aims of research and the broad time frame, but also specific, as regards the security of information, the data handling, the governance mechanisms, and the continuous information. The dynamic consent of the CHRIS study consists of two integrated components, the informed consent with dynamic options and the information material delivered through an ongoing multimedia, multilevel and culturally sensitive communication strategy.[8] The informed consent is an online interface, accessed for the first time and filled in at the study centre in Silandro, when prospective participants decide to enrol and participate in the CHRIS study. Through the online dynamic informed consent, participants express their preferences for each item of the consent. At any time afterwards, they can access their informed consent and visualize and change, if they wish so, their choices by entering MyCHRIS, a password-protected personal area of the website, which contains informed consent forms, clinical results, and further information on participation and data and samples use.

Prospective participants are invited through an invitation letter, which is sent at home. At the time of the initial consent (during the baseline), participants received all the relevant information through the brochure, which is integral part of the informed consent and whose aim is to explain all the aspects of the study. In order to participate in the first follow-up phase, invited participants receive a brochure about the follow-up study and sign a new dynamic informed consent at the study centre. Two brochures were issued so far: the first one, for the informed consent for participation in the CHRIS baseline, during the recruitment phase 2011-2018; the second one, for the informed consent of the first follow-up phase (from 2019). At the study centre, participants go through visual information material (videos, presentations), which explains the study and all the issues related to participation, and may ask the CHRIS study team questions and clarifications, if they wish so. The communication with study participants continues through the website, newsletters, emails, letters, text

[7] C. Pattaro, M. Gögele, D. Mascalzoni, R. Melotti, C. Schwienbacher, A. De Grandi, L. Foco, Y. D'Elia, B. Linder, C. Fuchsberger, C. Minelli, C. Egger, L. S. Kofink, S. Zanigni, T. Schäfer, M. F. Facheris, S. V. Smárason, A. Rossini, A. A. Hicks, H. Weiss, P.P. Pramstaller, *The Cooperative Health Research in South Tyrol (CHRIS) study: rationale, objectives, and preliminary results,* cit., 5-11; D. Noce, M. Gögele, C. Schwienbacher, G. Caprioli, A. De Grandi, L. Foco, S. Platzgummer, P.P. Pramstaller, C. Pattaro, *Sequential recruitment of study participants may inflate genetic heritability estimates*, in *Human Genetics*, 136, 6, 2017, 743-757; F. Murgia, R. Melotti, L. Foco, M. Gögele, V. Meraviglia, B. Motta, A. Steger, M. Toifl, D. Sinnecker, A. Müller, G. Merati, G. Schmidt, A. Rossini, P.P. Pramstaller, C. Pattaro, *Effects of smoking status, history and intensity on heart rate variability in the general population: The CHRIS study*, in *PLoS One*, 14, 4, 2019, 1-17; R. Melotti, R. Ruscheweyh, P.P. Pramstaller, A.A. Hicks, C. Pattaro, *Structural consistency of the pain sensitivity questionnaire in the Cooperative Health Research In South Tyrol (CHRIS) population-based study*, in *The Journal of Pain*, 19, 12, 2018, 1424-1434.

[8] For an overview of the dynamic consent of the CHRIS study: C. Pattaro, M. Gögele, D. Mascalzoni, R. Melotti, C. Schwienbacher, A. De Grandi, L. Foco, Y. D'Elia, B. Linder, C. Fuchsberger, C. Minelli, C. Egger, L. S. Kofink, S. Zanigni, T. Schäfer, M. F. Facheris, S. V. Smárason, A. Rossini, A. A. Hicks, H. Weiss, P. P. Pramstaller, *The Cooperative Health Research in South Tyrol (CHRIS) study: rationale, objectives, and preliminary results,* cit., 3-5. . Additionally, a manuscript aon an overall assessment of the CHRIS dynamic consent, focused on participation, trust, and communication during the recruitment phase is currently under review.

messages, and public conferences and press releases for the general public as well. All these different communication tools are meant to keep participants informed and updated about the development of the study, the achievements and outcomes of the research, and are used also to promote engagement and participation, and a trust-based relationship with participants. In the dynamic consent, an ongoing and continuous communication plays a key role in the informed consent process, and for the consent to be meaningfully informed through time.

With the beginning of the first follow-up phase, changes in several aspects of the study were introduced and novel aspects were added in comparison with the baseline. For example, new types of data and additional clinical measurements are collected, and new strategies of data collection are employed (e.g., through smart devices and remote phenotyping). Furthermore, participants are asked to give the CHRIS study permission to access their medical records (exclusively for health data which are relevant for the aim of the CHRIS study), and to consent to the production and storage of immortalized cell lines for functional studies. The ongoing consent process, and the related communication to participants have been refined and clarified (see below in 3.). The information on the governance for the return of results has been furtherly improved with a nuanced model describing four genetic diseases: participants are provided with a description of four types of genetic results, which are associated with pathologies differing for severity, transmission, opportunities for therapy and prevention. This information is meant to clarify the meaning and the implications of getting to know own genetic results associated with pathologies, and to enable an informed decision on the return of result option in the informed consent. The CHRIS follow-up phase was approved by the ethics board of Azienda Sanitaria dell'Alto Adige, and the new aspects were addressed in the information material provided to participants, and, of course, in the dynamic consent of the follow-up phase. The use of dynamic consent allowed to address changes reflecting both the researchers' scientific needs, and participants' interests as well, with the result of facilitating the conduction of biomedical research in the long-run, and of adjusting and improving governance mechanisms and communication with participants.

In the following paragraphs, we describe the dynamic informed consent and the contents of the information material (mainly, the brochure, the presentation, and the videos) of the CHRIS follow-up phase,[9] in order to provide an overview of what participants are asked to consent to, and which information they receive for an informed decision about participation.

2.2.1. Dynamic informed consent for participation in the follow-up phase of the CHRIS study

The dynamic consent of the CHRIS follow-up phase includes three main sections: the consent for the participation in the CHRIS follow-up, the consent for the use of data and samples in research, and the consent to the use of the online platform MyCHRIS.

In the consent for participation, participants consent to the collection of biological samples, to go through a series of clinical and scientific examinations, to answers interviews and questionnaires on general health, lifestyle, medical history. Participants can choose the way they prefer to receive the

[9] Informed consent forms and brochures are available in the CHRIS study website at https://it.chris.eurac.edu/download/ (last visited 19/01/2021)

results of the clinical examinations[10] (at the CHRIS centre, sent at home, through MyCHRIS or the smartphone application).

In the consent for data and samples use, participants consent to long-term storage of data and samples in the biobank (codified, non-identifiable data and samples) for the research purposes of the CHRIS study. Then, participants can choose through different options for some questions where they can approve or refuse. These are data sharing, functional studies with immortalized cell lines, access to medical records, use of data and samples in case of incapacity and death, return of results, and re-contact for participation in further studies. The choices made are changeable by using MyCHRIS or through other authorized channels (see below). In this way, participants are enabled to choose and modify the extent of their participation in research over time. Participants can withdraw from the study by contacting the CHRIS centre by phone or email.

My CHRIS is the core of the control by patients. Participants are asked to consent to the use of the dynamic consent online platform MyCHRIS, where they can legally modify the choices made in the consent, consent to new studies, and withdraw the consent. Through MyCHRIS, it is also possible to receive information on the studies, to participate in sub-studies remotely by filling-in questionnaires, and to access own clinical results. By using username and password, MyCHRIS is accessible through the CHRIS website or through the relative smartphone application. A one-time password sent through email or text message is needed in addition, as a system of two-factor authentication, for all those actions that imply an active decision by the participant to exert own will and rights, and that will have an impact in participation, such as to consent to new sub-studies, to modify the consent already given, to modify the password, to download own clinical records, to request the modification of personal data and contact information.

Participants are kept updated and, for studies whose aim does not fall within the areas covered by the general consent, participants will receive a notification and/or a consent request (opt-in or opt-out, as decided by the ethics board). For this reason, it is very important that participants express their preference on the way of communication with the CHRIS team: they are asked to choose at least two ways of re-contact among email, text message, CHRIS application, and phone, or, if the none of the other options are feasible, mail. In the consent form, participants are informed that all the changes made through MyCHRIS have legal value. Participants can also decide not to accept to the use of MyCHRIS as tool for consent management. In this situation, all the changes can occur via letter, signed by the participant and accompanied by a copy of a valid identity document, and all the relevant information about further studies will be given via phone or mail.

2.2.2. Information for an informed decision on participation in research

After the end of the recruitment and before re-inviting participants to the follow-up, all CHRIS participants received a newsletter, which informed them about the outcomes achieved during the recruitment phase and about the upcoming first follow-up phase. Information on the CHRIS study and on the follow-up phase were made available also in the CHRIS webpage. Participants that were progressively re-called for the follow-up appointment received an invitation letter, which briefly describes

[10] Participants are informed and aware that only specific measurements with clinical value (listed in the informed consent and in the brochure) are returned.

the study. If the invited CHRIS participants agree to participate, they can fix an appointment at the study centre, where they will proceed with the visit and the data and samples collection, after consenting to participate. Before the appointment, the participant is sent a brochure (sent at home through mail, paper version), which provides the information about participation, including all the changes introduced in the follow-up phase, participant rights, and governance. Both the newsletter and the brochure are available also in electronic version.

Through the brochure for the follow-up phase, participants are informed on the CHRIS study and its aim, about the benefits and risks of participation in research, and the ways of re-contact and interaction with participants. The clinical measurements and the other types of data collected are described. Participants are informed on which clinical measurements will be returned (that is, those of clinical value) and which ones will not be returned. They are also informed that individual genetic research results will not be communicated, with the exception of those that have relevance for the health of the participant or for the participant's family, upon decision of the ethics board and involvement of genetic counsellors, according to the international guidelines on medically actionable genes, and according to the participant's right to know or not to know. Four conditions (malignant hyperthermia, Parkinson's disease, breast cancer, and Huntington's disease) are described as model for the typology of genetic research results, and the mechanisms in place for the return of results are explained. Participants are informed about their rights and about other legal aspects (dynamic consent, consent to sub-studies, flow of information supporting the consent and on the development of the study, use of the dynamic consent platform MyCHRIS, withdraw). Furthermore, participants receive all the information on data and samples treatment and storage (access to medical records, privacy and protection, governance for data sharing, storage in the biobank, dissemination of research results, oversight bodies).

The information material paired with the informed consent of the CHRIS study includes also a voice-over presentation, which summarizes the study in the same line as the brochure, and videos about the four genetic diseases.

3. Data sharing and ongoing consent in the CHRIS dynamic consent: broad consent within strong governance, oversight mechanisms, and ongoing information

Data Sharing and ongoing consent are good examples to show how the CHRIS dynamic consent works in practice. We decided to focus on these aspects in order to show how dynamic consent allows participants to exert their autonomy by making individual choices about the extent of their participation. This occurs within established mechanisms of oversight and governance. By consenting to participate in research within the broad aim of the CHRIS study, participants consent to a governance structure which they are made aware of through the information material and the informed consent. The information that participants receive about the development of the study, about the sub-studies, and the governance mechanisms for further participation in research frames the broad research aim within the defined boundaries of the specific information provided. Through the samples illustrated in this section, we want to clarify how the dynamic consent can be used in practice, and thus rectify some of the misconceptions and misunderstandings about its use and potentialities.

CHRIS data and research results can be shared with other institutions and researchers, to be used for research purposes. Institutions and researchers interested in accessing the CHRIS repository must submit a formal request to the access committee, which oversees data and samples access.[11] The access committee is an internal body composed of multidisciplinary experts, which evaluates applications for data and samples access, by considering the scientific validity of the submitted project, its conformity to the scientific aims of the CHRIS study, and the compliance with ethical and legal regulations in place in the CHRIS study. Additionally, the projects that will make use of CHRIS data and samples must be approved by an ethics board. After approval of the access committee, access to data and samples occurs only after a contract (data/material transfer agreement, DTA/MTA) is signed. This contract establishes that the data are used according to the General Data Protection Regulation 2016/679 (GDPR).[12] As safety measure, only codified and non-identifiable data are shared. Furthermore, also the dissemination of results obtained by using the CHRIS data or samples must be approved by the access committee. How are the CHRIS participants given choices within the data sharing process? In the informed consent, they can agree or not to sharing, and they can express their preferences on the extent of it: they can specifically agree (or not) to sharing with partner institutions that are bound through a DTA/MTA, and/or with larger scientific institutions (such as databases and consortia) which allow maximization of data usage. Participants are informed that the legal regulations might differ if data sharing occurs with institutions which are not under the GDPR. In MyCHRIS, participants are informed about the projects that their data are being shared in, they are updated about the research development through the webpage, and/or other apt communication channels. The flow of information will allow them to be aware of what happens with their data and samples, and of how the governance works, and, therefore, to retain control of their data and samples by having the possibility of changing the degree of their participation regarding data sharing in the dynamic informed consent platform. In this way, they receive all the information that put their individual choice on participation within the research process and the oversight mechanisms in place, while making an autonomous decision on participation that reflects their values through time.

Dynamic consent has been criticized for the risk of impairing the governance of the biobank.[13] However, by describing how the data sharing is handled in the CHRIS study through dynamic consent, it is evident that the authority of deciding on scientific value of research, and compliance to ethical and legal standard stays in the hands of experts and oversight bodies, such as researchers, the access committee and the ethics board. Therefore, for the way in which dynamic consent is implemented in the CHRIS study, the authority of the governance and of the oversight bodies is neither weakened nor transferred to participants.[14]

[11] The form and guidelines for application for data and samples access in the CHRIS study are available in the Eurac website at http://www.eurac.edu/en/research/health/biomed/projects/Pages/CHRIS.aspx (last visited 12/01/2021).

[12] GDPR: https://eur-lex.europa.eu/eli/reg/2016/679/oj (last visited 18/01/2021).

[13] A. SOULIER, *Reconsidering dynamic consent in biobanking: Ethical and political consequences of transforming research participants into ICT users*, in *IEEE Technology and Society Magazine*, 38, 2, 2019, 62-70.

[14] We want to add further nuance on this issue, by suggesting that research participants can play an active role within the research process and the governance structure by providing inputs and insights on research from their perspective. In fact, CHRIS participants have been actually involved in policy design and recommendations

As the CHRIS study is designed as prospective and longitudinal, and the CHRIS biobank is meant as a dynamic resource for research, CHRIS participants' data and samples are foreseen to be used for several future studies. However, these possible future studies may not be explicitly described, defined or known at the time of the initial consent. Through dynamic consent, CHRIS participants agree to an initial broad consent for the collection and use of data and samples within the scientific areas of interest of the CHRIS study and the limits of the informed consent. In order to keep participants aware of the development of the study and of the use of their data and samples in sub-studies that were already described at the time of the initial consent and therefore within the CHRIS objectives, participants are informed through MyCHRIS and through the other communication channels of the CHRIS study (webpage, newsletter). However, in order to participate in other further studies, they may be asked to re-consent. For new studies within the main aim of the CHRIS study, but that were not enough described or defined at the time of initial consent, participants are asked to re-consent through opt-in or to opt-out, following an ad hoc communication through the tools preferred by the participant (email, text message, phone, mail). The opt-in strategy is for specific projects which were not foreseen in the description of the CHRIS study, while the opt-out strategy applies to specific projects within the aim of the CHRIS study and already described, but not detailed enough at the time of the initial consent. With the opt-in strategy, participants are asked to provide an "active" consent, while in the opt-out they are included in the study unless they actively refuse to participate ("passive" consent). In both the opt-in and the opt-out strategy, an ad hoc communication (information material through email and MyCHRIS, possibility to ask the CHRIS study team clarifications and questions) supports the active or passive consent request. The strategy for such studies is summarized in Table 1.[15]

Already described studies, whose aim is within the areas of the CHRIS study and the limit of the informed consent	Specific projects not foreseen in the description of the CHRIS study	Studies that were described but not detailed enough
Communication through MyCHRIS and the other CHRIS communication channels No re-consent	Ad hoc communication with specific information Opt-in ("active consent") through MyCHRIS	Ad hoc communication with specific information Opt-out ("passive consent") through MyCHRIS

Table 1. Ongoing consent in the CHRIS study: consent and respective communication strategy

development on recall-by-genotype and return of results. With this, we mean that their views on recall-by-genotype and on return of results were investigated and explored through empirical studies, and the results of these studies (one manuscript under review and one manuscript in preparation) will contribute to the understanding of participants' wishes on participation in genetic research and their views on the possible communication of genetic results, and to further develop and improve the policy on such issue. Therefore, CHRIS participants' contribution in the biobank policy and governance should be understood in the context of a dialogue with the researchers and the experts in charge of the governance.

[15] The table is an adaptation from the CHRIS follow-up brochure (p. 27), which is available at https://it.chris.eurac.edu/download/ (last visited 31/01/2021).

For new studies whose aim is beyond the one that participant consented to and where a new participation (e.g., new data and samples collection) is asked, a new specific informed consent, accompanied by specific informative material, is asked. The ethics board deliberates for each approved project which type of consent strategy is needed. Through these differential re-consent strategies and the ongoing consent process, the broad consent regarding the research scope is situated within specific and defined conditions.

The governance on communication, the oversight on the scientific aim and the ongoing consent process provide the level of specificity that is required as an ethical and legal requirement in research, and especially in broad research endeavours such as biobanking. As in the data-sharing situation, the flow of information allows participants to make informed decision on their participation in research through time and to exert their rights through the dynamic consent tools. So, it is a misconception that, when participating in research through dynamic consent, participants will be asked to re-consent all the time for every single new research project and that the ethical assessment will be weakened,[16] because as shown by the case of the CHRIS study, the ongoing consent process is held within a specific governance.

This includes the ethics board, which defines the approach to re-consent or notification that should be applied, and an ongoing information policy and communication structure, meant to notify and update participants about the research projects that are conducted within the biobank so that they receive all the information for the consent to be properly informed.

On a different note, even though dynamic consent relies on information technology and communication strategies broadly embedded within digital and electronic platform and devices, this does not exclude alternative ways of communication. More traditional communication tools (such as phone or paper-based material, e.g. mail, newsletter, etc.) are in place as an alternative to guarantee that participants are not excluded as a result of the digital divide.[17] In fact, in the CHRIS study, participants who decide that they prefer not to use MyCHRIS, the online platform of the dynamic consent, are offered other strategies to be informed and contacted about research studies, and to exert their rights as research participants.

4. Conclusion

With this paper we wanted to showcase how the dynamic consent model was implemented in the frame of the CHRIS study, a population study conducted in South Tyrol since 2011. The dynamic consent of the CHRIS study can be intended as a broad consent (as regards the aims of research and the long-term storage and use of data and samples) within strong oversight and governance mechanisms, paired with an ongoing communication with participants. By discussing the mechanisms of data sharing and ongoing consent in the CHRIS follow-up phase, we took the chance to address some of what we considered as misconceptions regarding dynamic consent and its actual implementation in

[16] K.S. Steinsbekk, B.K. Myskja, B. Solberg, *Broad consent versus dynamic consent in biobank research: Is passive participation an ethical problem?*, in *European Journal of Human Genetics,* 21, 2013, 898-901.

[17] M. Prictor, H.J.A. Teare, J. Kaye, *Equitable Participation in Biobanks: The risks and benefits of a "Dynamic Consent" approach*, in *Frontiers in Public Health*, 6, 253, 2018, 4.

practical terms. Given this premise, our experience with the dynamic consent of the CHRIS study becomes even more relevant in the context of the current debate on informed consent in biobanking research. We believe that it is important to share our experience in the dynamic consent of the CHRIS study with the scientific community, because it can serve as an example of practical implementation of a model suitable for long-term genomics research and biobanking research.

Special issue

A New Era for Biotech Patents? Empirical and Theoretical Considerations on the current Patent Dilemma

Nicola Lucchi*

ABSTRACT: During the last two decades, the genomics revolution has contributed enormously to the development of novel research approaches in the field of biological sciences. We have seen the development of new biotechnological tools capable of modifying organisms in order to perform specific tasks designing and assembling novel biological components. All these scientific and biotechnological innovations present also a substantial challenge for the law and especially for intellectual property rights. In particular – as a consequence of relevant patent case law in the United States and Europe affecting precision medicine – the debate on the possible future of biotech patents gained increasing momentum. Considering this multifaceted scenario, the purpose of this article is to analyse the impact of various judicial decisions with respect to patents subject-matter eligibility and the prosecution of biotech-related patent applications.

KEYWORDS: Gene patents; genetic engineering; biotechnology; patents; CoViD-19

SUMMARY: 1. Introduction – 2. The old era of Biotech Patents – 3. A paradigm shift on future biotech patents? – 4. The problem of Intellectual Assets in the Life Sciences Industry – 5. The impact of court decisions for the Life Sciences Industry: Myriad and Mayo – 6. The impact of court decisions for the Life Sciences Industry: Brüstle – 7. Conclusion.

1. Introduction

Legal questions concerning the interpretation and application of patent laws to the scientific area have started to be challenged more frequently also in courts in different countries. In particular, the effects and consequences of these controversial judgments have been intensively discussed both in academia and policy circles. Courts and scholars have reopened and rekindled the debate over the benefits and risks associated with patent protection to diagnostic methods and other scientific research techniques. For the most part, these discussions are focused on patent applications with the aim to determine the patentability of a particular invention related to human DNA sequences or stem cells developed in vitro and used for scientific research.

In recent years – especially in the United States – there have been a series of highly controversial court decisions involving subject-matter eligibility for patents in genes.[1] At the same time in Europe,

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ISSN 2284-4503

the patentability of inventions relating to human embryonic stem cells has raised morality-related issues culminated in a series of decisions of the Court of Justice of the European Union in an attempt to define uses of human embryos for industrial and commercial purposes.

In particular, these decisions have triggered a series of questions about the future of patentability in the field of innovative diagnostic methods, biomarker and personalized medicine "with potentially profound implications for the biotech industry".[2] The question here is whether there is empirical evidence that these judgments actually had a negative impact in the field of precision medicine and in DNA-based diagnostics depriving certainty in patent law. This is a topic on which – as far as I know – very few empirical studies have been conducted to date and it is thus a particularly interesting field of investigation.[3] This debate has found its way into various scholarly works, without finding yet a pertinent solution for the questions raised.[4]

In the following pages, we will try to clarify the possible and actual negative effects arising from these disputes by reviewing some recent studies that have empirically evaluated how these cases actually affect the future of biotech patents, including patent filing and prosecution.[5]

2. The old era of Biotech Patents

The exceptionally rapid technological progress in the field of biotechnology has resulted in more appropriate legislative and judicial responses to the evolving regulatory regime especially with regard

[1] See *Mayo Collaborative Servs. v. Prometheus Labs*., Inc., 132 S. Ct. 1289 2012; *Association for Molecular Pathology v. Myriad Genetics*, Inc., 133 S. Ct. 2107 2013.

[2] M. ABOY et al., *Mayo's impact on patent applications related to biotechnology, diagnostics and personalized medicine*, in *Nature Biotechnology,* 37, 2019, 513, 513.

[3] E.J. HAANES, J.M. CANAVES, *Stealing Fire: A Retrospective Survey of Biotech Patent Claims in the Wake of Mayo v. Prometheus*, in *Nature Biotechnology*, 30, 2012, 758.

[4] See e.g. J. LIDDICOAT et al., *The Effects of Myriad and Mayo on Molecular-Test Development in the United States and Europe: Interviews from the Frontline*, in *Vanderbilt Journal of Entertainment and Technology Law,* 22, 2020, 785; M. ABOY et al., *One year after Vanda, are diagnostics patents transforming into methods of treatment to overcome Mayo-based rejections?*, in *Nature Biotechnology,* 38, 2020, 279; J. PILA, *Adapting the ordre public and morality exclusion of European patent law to accommodate emerging technologies*, in *Nature Biotechnology,* 38, 2020, 555; M. ABOY et al., *Was the Myriad Decision a 'Surgical Strike' on Isolated DNA Patents, or Does it Have Wider Impacts?,* in *Nature Biotechnology*, 36, 2018, 1146; M. ABOY et al., *How does emerging patent case law in the US and Europe affect precision medicine?*, in *Nature Biotechnology,* 37, 2019, 1118; M. ABOY et al., *After Myriad, what makes a gene patent claim 'markedly different' from nature?*, in *Nature Biotechnology,* 35, 2017, 820; M. ABOY et al*., Myriad's impact on gene patents*, in *Nature Biotechnology,* 34, 2016, 1119; A. MAHALATCHIMY et al., *The impact of European embryonic stem cell patent decisions on research strategies*, in *Nature Biotechnology,* 33, 2015, 41.

[5] In particular, M. ABOY et al., *After Myriad , What Makes a Gene Patent Claim Markedly Different' from Nature?,* in *Nature Biotechnology,* 35, 2017, 820; M. ABOY et al., *Mayo's Impact on Patent Applications Related to Biotechnology, Diagnostics and Personalized Medicine*, in *Nature Biotechnology,* 37, 2019, 513; M. ABOY et al., *Myriad's Impact on Gene Patents*, in *Nature Biotechnology*, 34, 2016, 1119; M. ABOY et al., *Was the Myriad Decision a "Surgical Strike" on Isolated DNA Patents, or Does It Have Wider Impacts?*, in *Nature Biotechnology*, 36, 2018, 1146; G.D. GRAFF et al., *Not Quite a Myriad of Gene Patents*, in *Nature Biotechnology,* 31, 404, 2013; E.J. HAANES, J.M. CANAVES, *Stealing Fire: A Retrospective Survey of Biotech Patent Claims in the Wake of Mayo v. Prometheus,* in *Nature Biotechnology,* 30, 2012, 758.

ISSN 2284-4503

to intellectual property rights, trying to balance the interests of both rights-holders and individuals.6 The key to the success of the biotechnology industry was – *in fact* – the result of numerous supporting and "favourable government policies toward the sector in combination with a liberalized regime for the governance of biomedical research".[7] Debates and "concerns about the need to regulate the disruptive potential of biological manipulation were apparent almost from the moment when genetic engineering became feasible in the 1970s".[8] The emergence of all these issues and concerns set the basis for a new discussion on how to regulate the use, access, distribution, and appropriation of essential public knowledge assets in the life sciences.

We are presently living within one of the clearest proofs of the failure of this model: the current COVID-19 pandemic is showing us all the contradictions of the current patent system. If democratic States may legitimately interfere with certain fundamental rights through limitations or derogations in order to contain the COVID-19 pandemic, it is certainly incomprehensible how – also in respect of a proper balance of rights – it is not equally reasonable to temporarily derogate from patent protection in order to guarantee safe and equitable access to life-saving drugs and vaccines.9 Today, interests antagonistic to the right to health, freedom of movements, freedom of enterprise and access to scientific knowledge are emerging distinctly, requiring a careful balance between public and private interests. A significant proportion of DNA-related patents are owned by companies with a direct interest in commercialising genetic diagnostics. These patents often claim whole genes, gene fragments, cDNA, mutations, nucleic acids and DNA sequences "which hybridise under stringent conditions to the primary DNA sequences just to stop competitors taking advantage of redundancies in genetic code".[10]

Considering this scenario, the purpose of this investigation is to briefly outline the past and take a closer look at the future of biotech patents. In particular, the reason behind this analysis is to assess if – as a result of some court rulings – there are actual data for believing that the number of gene patents is declining and will continue to decline as well as whether it is a good or bad thing.[11] It was more than a decade ago when some US Supreme Court decisions (hereinafter *Mayo*[12], *Myriad*[13] and *Alice*[14]) have – in fact – completely changed US patent eligibility criteria for biotechnology reversing more than 30 years of case law questioning the enforceability and validity of patents on naturally occurring genetic material (even if isolated). Specifically, in *Myriad*, the Supreme Court held that

[6] See N. LUCCHI, *The Impact of Science and Technology on the Rights of the Individual*, 2016, 67.

[7] See S. VALLAS, D.L. KLEINMAN, D. BISCOTTI, *Political Structures and the Making of U.S. Biotechnology, in State of Innovation: The U.S. Government's Role in Technology Development*, in F. BLOCK, M. KELLER (eds.), *State of Innovation. The U.S. Government's Role in Technology Development,* 2011, 57.

[8] See S. JASANOFF, *Rewriting Life, Reframing Rights*, in ID. (ed.), *Reframing Rights: Bioconstitutionalism in the Genetic Age,* 2011, 1, 9.

[9] See E. BONADIO, A. BALDINI, *COVID-19, Patents and the Never-Ending Tension between Proprietary Rights and the Protection of Public Health*, in *European Journal of Risk Regulation,* 11, 2020, 390 (illustrating the patent race in the context of the Covid-19 pandemic).

[10] See K. LIDDELL et al., *Patents as incentives for translational and evaluative research: the case of genetic tests and their improved clinical performance*, in *Intellectual Property Quarterly,* 3, 2008, 286, 296.

[11] See e.g. M.M. HOPKINS et al., *DNA Patenting: The End of an Era?,* in *Nature Biotechnology,* 25, 2007, 185.

[12] *Mayo Collaborative Servs. v. Prometheus Labs*., Inc., 132 S. Ct. 1289, 2012.

[13] *Association for Molecular Pathology v. Myriad Genetics*, Inc., 133 S. Ct. 2107, 2013.

[14] *Alice Corporation v. CLS Bank International*, 132 S. Ct. 2347, 2014.

naturally-occurring DNA is not eligible for patenting just because it has been isolated from its natural state. Naturally occurring DNA remains a "product of nature" even after isolation and, therefore, falls under the "laws of nature" exception to patent eligibility. In other words, isolation of naturally-occurring DNA is not enough for the naturally-occurring DNA to be considered man-made. At the same time in *Mayo*, the US Supreme Court held that certain diagnostic methods are not patentable because they involved standard conventional steps. In particular, the Court found that the procedure by which a drug is chemically converted in the body to a metabolite must be considered as a natural process, and so the relationship between the quantities of metabolites and the efficacy of drug dose is a law of nature. In other words, simply observing such a naturally-occurring relationship – without adding anything else – cannot be considered enough to transform the concept of the claims into a patentable invention. Finally, in *Alice*, the US Supreme Court entirely reaffirmed the Mayo framework applying it to claims directed to computer-implemented process, computer system, and computer readable medium for mitigating settlement risk. In particular, the court determined that the method claims, which merely required generic computer implementation, are not sufficient to transform the abstract idea into a patent-eligible invention.[15]

In Europe, biotech patent disputes were raised by the landmark case of *Brüstle vs Greenpeace*, where the Court of Justice of the European Union was confronted with moral and legal dimensions of embryonic stem cells research. In this decision, the CJEU held that processes that require the use of stem cells gathered from a human embryo involving the destruction of that embryo cannot be subject to a patent. In particular, the Court recognized that "any human ovum after fertilization, any non-fertilized human ovum into which the cell nucleus from a mature human cell has been transplanted, any non-fertilized human ovum whose division and further development have been stimulated by parthenogenesis constitute a 'human embryo'"[16] thus constituting an unpatentable subject matter within the meaning of Article 6(2)(c) of the Biotech Directive.[17] Just two years later – surprisingly enough – the CJEU then allowed the patenting of human stem cells derived from unfertilized ova.[18] And finally, in 2005 the CJEU upheld the interpretation established in the *Brüstle* case that the concept of the human embryo within the context of the biotech directive must be understood in a broad sense.[19]

All these cases illustrate the difficulties of courts in reviewing scientific and technological issues as well as their huge power in creating a significant overlay of doctrine that may change completely the meaning or the interpretation of statute law. In particular, the role of the judiciary seems to have become decisive in "patent law assessment of legal subject matters".[20] In addition, these cases show how the granting of patents in the biotech sector may imply – at least in Europe – heterogeneous

[15] *Alice Corporation v. CLS Bank International*, 132 S. Ct. 2347, 2014.

[16] See *Oliver Brüstle v Greenpeace* eV (C-34/10), 2012. 1 C.M.L.R. 41, at § 38.

[17] Directive 98/44/EC of the European Parliament and of the Council of 6 July 1998 on the legal protection of biotechnological inventions, OJ EC L 213, 30 July 1998, 13-21.

[18] Case C-364/13, *Int'l Stem Cell Corp. v. Comptroller Gen. of Patens, Designs and Trade Marks*, ECLI:EU:C:2014:2451

[19] Case C- 456/ 03, *Commission v. Italy*, 2005 E.C.R. I-5355, recital 24.

[20] See F. MOLNÁR-GÁBOR, *Science, ethics, and patents. Ethically-motivated barriers for the patenting of the results of human embryonic stem cell research*, in C. HAUSKELLER et al. (eds), *The Matrix of Stem Cell Research: an Approach of Rethinking Science in Society*, 2019, 50.

and controversial ethical or moral elements that are difficult to harmonize at both national and European level. In contrast with the European Patent Convention and the EU Biotech directive, the United States patent system does not explicitly outline what categories of inventions or discoveries are placed outside the umbrella of patentability.[21] It is thus left to the Courts to define – time by time – where the boundary of patentability lies, and they are bound only by the provisions of the legislation. At the same time, the "public morality argument" as well as the phenomenon of the "ethicalization of law" are not particularly evident within American patent law.[22]

The comprehensive new approach resulting from these different decisions narrowed "de facto" patent-eligible protection over living organisms and created significant ambiguity among both patent applicants and patent examiners. In order to mitigate this confusion, the United States Patent and Trademark Office (USPTO) arranged a series of guidance on subject matter eligibility. These new challenges faced by biotechnology innovators have possibly fuelled the debate over new effective instruments for supporting innovations in the biotech sector: this discussion – which also came to reconsider the idea of using alternative form of intellectual property in order to protect engineered nucleic acid sequences[23] – is probably demonstrating the need to draw a completely new set of principles for biotech patents.

3 A paradigm shift on future biotech patents?

The standard approach of the Trilateral Patent Offices [i.e. the United States Patent and Trademark Office (USPTO), the Japan Patent Office (JPO) and the European Patent Office (EPO)] with respect to

[21] See T. Minssen, D. Nilson, *Standing on shaky ground: US patent-eligibility of isolated DNA and genetic diagnostics after AMP v USPTO* – Part I, in *Queen Mary Journal of Intellectual Property*, 1, 2011, 223 - 224.

[22] See R. A. Spinello, *A Defense of Intellectual Property Rights*, in *Elgar*, 2009, 68 (noting that that the public morality aspect is feeble in American patent law).

[23] See D.L. Burk, *DNA Copyright in the Administrative State*, in *UC Davis Law Review*, 51, 2018, 1297; N. Lucchi, *Genetic Copyright: An Alternative Method for Protecting and Using Essential Public Knowledge Assets?,* in European intellectual property review, 40, 2018, 766; R. Neethu*, Rekindling the debate on genetic copyright in Europe in the era of biobanks and synthetic biology*, in European intellectual property review, 40, 2018 172; C.M. Holman, *Charting the Contours of a Copyright Regime Optimized for Engineered Genetic Code*, in *Oklahoma Law Review,* 69, 2017, 399; C.M. Holman, *Copyright for Engineered DNA: An Idea Whose Time Has Come*, in *West Virginia Law Review,* 113, 2011, 699; C.M. Holman, *Developments in synthetic biology are altering the IP imperatives of biotechnology*, in *Vanderbilt Journal of Entertainment and Technology Law,* 17, 2015, 385; C.M. Holman, *Copyright for Engineered DNA*, in *GQ Life Sciences*, available at https://bit.ly/3dJnRBe, (last visited 27/04/2021); A. W. Torrance, *Synthesizing Law for Synthetic Biology*, in *Minnesota Journal of Law, Science & Technology,* 11, 2010, 629; M.D. Murray, *Post-Myriad Genetics Copyright of Synthetic Biology and Living Media*, in *Oklahoma Law Review,* 10, 2014, 71; D. Walker, *Patent Protection or Copyright for Nucleic Acid Sequences?*, in *Licensing Journal*, 36, 2016, 1; J. Roig, *Can DNA Be Speech?*, in *Cardozo Law's Arts & Entertainment Law Journal,* 34, 2016, 163; J.J. Zhuang, *Copyright: Better Fitting Genes*, in *Journal of the Patent and Trademark Office Society,* 97, 2015, 442; J.N. Michelotti, *Genes as Intellectual Property*, in *Michigan State University Journal of Medicine and Law*, 11, 2007, 71; T. Chen, *Can a Biological Sequence Be Copyrighted*, in *Intellectual Property and Technology Law Journal,* 19, 2007, 1.

patents on biological subject matter is to grant ownership rights only for isolated and purified gene sequences with a demonstrated specific utility.[24]

A clear dividing line between patentable subject matter and non-patentable products of nature was established for the first time by the United States Supreme Court in Diamond v. Chakrabarty.[25] This historical court decision – in combination with the judgement in Moore v. Regent of University of California[26] – not only significantly impacted the U.S. patent system, but provided a new lens to look at how genetic resources can be used and privatized. A few years later, the USPTO, EPO, and JPO released a joint policy statement claiming that "purified natural products are not regarded as products of nature or discoveries because they do not, in fact, exist in nature in an isolated form. Rather, they are regarded for patent purposes as biologically active substances or chemical compounds and eligible for patenting on the same basis as other chemical compounds".[27]

The aim of this statement was to identify the relevant global planning policy regarding patentability of genetic material: in particular, it was specified that a purified natural substance was to be considered patentable if the "purification" results in a compound with such distinct characteristics that it becomes a new product commercially or therapeutically valuable. In the process of isolation and purification of genetic materials, it is – in fact – possible to obtain the partition of different compounds from a biological cell. However, it should also be pointed out that various criticisms have been made related to the above interpretation. In particular, it has been stressed that even if genetic materials are purified and isolated, the core elements of such substances – which are the "useful" and exploitable information – "are naturally occurring, not created by the person who isolates and purifies the material".[28] In addition, purified and isolated genetic sequences are "structurally similar or identical to the form that exists in nature".[29]The main point of this interpretation is that patents for biotech innovations are simply based and limited only on the ability of the individuals drafting the claim.[30]

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[24] See M.J. HOWLETT, A.F. CHRISTIE, *An Analysis of the Approach of the European, Japanese and United States Patent Offices to Patenting Partial DNA Sequences* (ESTs), in *Int'l Rev. Indus. Prop. & Copyright L.,* 34, 2003, 581; L.G. RESTAINO et al., *Patenting DNA-Related Inventions in the European Union, United States and Japan: A Trilateral Approach or a Study in Contrast?*, in *UCLA Journal of Law and Technology*, 2003, 2. See also A. REESE, B. OPESKIN, *Current Issues in Gene Patenting*, in I. FRECKELTON, K. PETERSEN (eds.), *Disputes and Dilemmas in Health Law*, 2006, 277, 280; N. LUCCHI, *Understanding genetic information as a commons: from bioprospecting to personalized medicine*, in *International Journal of the Commons*, 7, 2013, 313.

[25] *Diamond v. Chakrabarty*, 447 U.S. 303 (1980). In this landmark decision, the United States Supreme Court held that a live and human-engineered microorganism can be considered a patentable subject matter under Section 1010 of the *United States Patent Act*. According to the rule of this decision, patents can be issued on "anything under the sun that is made by man".

[26] *Moore v. Regents of the University of California*, 793 P.2d 479 (Cal. S. Ct) (1990).

[27] See 1988 Joint Statement of USPTO, EPO and JPO; *Comparative Study of Patent Practices in the Field of Biotechnology Related Mainly to Microbiological Inventions*, in *Biotechnology L. Rep*, 7, 1998, 159, 163; Nuffield Council of Bioethics Discussion Paper, *The Ethics of Patenting DNA*, 2002, 26, 3.14.

[28] See Australian Law Reform Commission, Genes and Ingenuity: Gene Patenting and Human Health, Report 99. Sydney: Australian Commonwealth, 2004, at 126. Available at: https://bit.ly/3dTzghW (last visited 27/04/2021).

[29] *Ibidem*

[30] See D. ROBINSON, N. MEDLOCK, *Diamond v. Chakrabarty: A Retrospective on 25 Years of Biotech Patents*, in *Intellectual Property and Technology Law Journal,* 17, 2005, 12, 14.

In more recent years, the patenting of genes and gene fragments has put under debate one of the fundamental principles of the patent law: the requirement of novelty.[31] Consequently, the debate on the status as patentable subject matter has suddenly become topical again. Indeed, many questions surround this issue: "as DNA has existed well before the gene discoverer arrived, how can these molecules be novel?"[32].The answer, as it has been suggested, "is that the actual molecule produced and claimed by the gene discoverer is new in a strict sense of the word".[33] More specifically, "gene sequences exist naturally as part of a much bigger molecule" and "there is no doubt that this much bigger molecule would be unpatentable".[34] But, on the other hand, the gene discoverer's thesis is that "purified and isolated gene sequences are distinct from the overall DNA molecule".[35] This is also the thesis formulated by one of the first U.S. patent infringement litigations involving a gene patent. In Amgen, Inc. v. Chugai Pharm. Co. Ltd., the district court ruled that the patent in suit was to be regarded as valid because the invention "is not as plaintiff argues the DNA sequence encoding human erythropoietin since that is a non-patentable natural phenomenon "free to all men and reserved exclusively to none". Rather, the invention as claimed in claim two of the patent is the "purified and isolated" DNA sequence encoding erythropoietin."[36]

In order to be patentable under the United States and European law, an invention must meet three basic requirements:[37] (i) novelty; (ii) inventive step (non-obviousness in the US); and (iii) industrial application (utility in the US). These statutory limits provide the basic and general requirements that must be satisfied in order to obtain a patent. However, patents on DNA and human genes raise the question of where to draw the line between patentable and non-patentable inventions. In fact, the patenting of genes and gene fragments appears to challenge the novelty requirement. At the same time, there is a growing sensitivity to ethics in patent law.[38] This sensitivity is even more evident in the biotech sector where the structure of the patent system seems to require a more careful assessment of all the possible conflicting rights.

The patent dilemma in the biotech sector is also challenged by a regulatory framework built for a more conventional setting. For example, there are increasing policy and academic discussions about the ethical and social issues raised by owning, managing and using essential public knowledge assets

[31] See e.g. J.J. DOLL, *The Patenting of DNA*, in *Science,* 280, 1998, 689; D.J. KEVLES, A. BERKOWITZ, *The Gene Patenting Controversy: A Convergence of Law, Economic Interests, and Ethics*, in *Brooklyn Law Review,* 67, 2001, 233. For a recent overview of the gene patenting controversies, see also L. LARRIMORE OUELLETTE, Access to Bio-Knowledge: *From Gene Patents to Biomedical Materials*, in *Stanford Technology Law Review,* 2010, available at https://stanford.io/2PrVWwl (last visited 27/04/2021).

[32] See O. LIIVAK, *Maintaining Competition in Copying: Narrowing the Scope of Gene Patents*, in *UC Davis Law Review*, 41, 2007, 177, fn 53.

[33] *Ibidem*.

[34] *Ibidem*.

[35] *Ibidem*.

[36] See *Amgen, Inc. v. Chugai Pharm. Co.,* 13 U.S.P.Q.2d (BNA) 1737, 1759 (D. Mass. 1989).

[37] See O. MILLS, *Biotechnological Inventions. Moral Restraints and Patent Law*, 2010, 2nd ed., 4; L. BENTLY, B. SHERMAN, *Intellectual Property Law*, 2018, 5th ed., 551.

[38] See e.g. judgment of the Court of Justice of the European Union in *Oliver Brüstle v Greenpeace* eV (C-34/10) [2012] 1 C.M.L.R. 41.

in the life sciences.[39] From the mere legal perspective, article 27 of TRIPs defines patentable subject matter expressly stating that patents must "be available for any inventions, whether products or processes, in all fields of technology, provided that they are new, involve an inventive step and are capable of industrial application" (the so called "Non-Discrimination Principle").[40] It means that genes, gene fragments and cell lines modified or altered by human effort can be patented if the inventor meets the general requirements of a patent.[41] Formally, States may refuse to recognize patents on their territory, but − up to now − very few countries have used this option. At the same time, the European Directive on the Legal Protection of Biotechnological Inventions stipulates that "elements isolated from the human body or otherwise produced by means of a technical process, including the sequence or partial sequence of a gene" may represent a patentable invention.[42] In particular, the Directive specifies that "biological material which is isolated from its natural environment or produced by means of a technical process is considered to be an invention even if this material previously occurred in nature".[43] In addition, the European Patent Convention (EPC) excludes the possibility to grant patents for "methods of treatment of the human or animal body by surgery or therapy and diagnostic methods practiced on the human body".[44] On this basis, the European Patent Office determined that "all methods practiced on the human or animal body which relate to the diagnosis or which are of value for the purposes of diagnosis" are precluded from being patented.[45] Nevertheless, biotech and life science-related inventions are in principle considered patentable under both the EPC and the Biotechnology Directive.[46] Specifically, the European Patent Convention unequivocally recognizes the patentability of biotech inventions in Rule 26(1) EPC.[47] In addition, Rule 27(a) EPC offers complementary details about the "patentable subject-matter" for life sciences inventions, specifying that "biotechnological inventions shall also be patentable if they concern biological material which is isolated from its natural environment or produced by means of a technical process even if it previously occurred in nature".[48] As a consequence – unlike in the Unites States where, after Myriad, isolated DNA sequences are no more eligible – biological material,

[39] D.M. GITTER, *International Conflicts Over Patenting Human DNA Sequences in the United States and the European Union: An Argument for Compulsory Licensing and a Fair-Use Exemption*, in *New York University Law Review*, 76, 2001, 1623, 1624.

[40] See Agreement on Trade-Related Aspects of Intellectual Property Rights, Apr. 15, 1994, art. 27, Marrakesh Agreement Establishing the World Trade Organization, Annex 1C, 33 I.L.M. 1125, 1994, [hereinafter TRIPS].

[41] L.B. ANDREWS, J. PARADISE, Essay, *Gene Patents: The Need for Bioethics Scrutiny and Legal Change*, in *Yale Journal of Health Policy, Law, and Ethics,* 5, 2005, 403, 404.

[42] Council Directive 98/44/EC, art. 5(2), 1998 O.J. (L 213) 13 (EC).

[43] *Id*., at art. 3(2).

[44] Convention on the Grant of European Patents, art. 53(c), Oct. 5, 1973, 13 I.L.M. 270 [hereinafter EPC].

[45] See decision T 964/99 (OJ EPO 2002, 4), starting from the interpretation set out in decision T 385/86 and decision T 964/99.

[46] See R.A. SPINELLO, M. BOTTIS, *A Defense of Intellectual Property Rights*, in *Elgar*, 2009, 64.

[47] Rule 26(1) of 5 October 1973 as adopted by decision of the Administrative Council of the European Patent Organisation of December 7, 2006 and as last amended by decision of the Administrative Council of the European Patent Organisation of October 26, 2010 [hereinafter Implementing Regulations]. The Rules cited are to the earlier version. On this, see G. MACCHIA, *Patentability Requirements of Biotech Inventions at the European Patent Office: Ethical Issues*, in *Biotech Innovations & Fundamental Rights*, R. BIN et al, eds, in *Springer*, 2011, 37.

[48] See Implementing Regulations, Rule 27(a).

including DNA sequences, which is isolated from its natural environment or produced with a technical process, is eligible for patent protection.[49]

4 The problem of Intellectual Assets in the Life Sciences Industry

Biotechnology and pharmaceutical corporations normally follow a conventional business model that is focused on a "closed innovation" scheme supporting a system that is completely enclosed by intellectual property rights. All ideas are internally generated and stay inside until the new product or innovation arrives on the market. In this context, it is relevant to consider how patent rights affect the process of "cumulative innovation".[50] Normally all inventions are based on previous knowledge and inventions: this means that innovation is cumulative because new innovations are grounded on previous innovations. The term "cumulative innovation" is commonly employed to describe a condition in which a second inventor uses previous knowledge protected by a granted patent in order to create a new innovation.[51] In other words, the second innovation would not be achievable without the contribution of the previous scientific knowledge. As a consequence, the second innovator is necessitated to obtain a license from the first innovator in order to use and exploit the new invention. The cumulative effect of innovation necessarily prompts serious concerns regarding the significance of dissemination of and access to scientific information.[52] Privately funded research in the life sciences is normally profit-oriented and patents are the primary strategy that firms use to protect their new ideas. In such specific circumstances, concerns arise because of the nature and extent of protection granted to patent holders. Patents – in fact – may play various roles in the knowledge-based economy. Under the current regulatory framework, patent holders have broad freedom in the exercise of their exclusive prerogatives.[53] Consequently, they are free to negotiate and set royalties, to accept, deny or unreasonably limit licensing requests, or again they may select specific licensees imposing licensing terms freely, as long as the terms and agreements do not override relevant regulations, such as competition or antitrust law.[54] Unfortunately, this scheme – even though designed to support private research – could also bring undesirable results. When patents are licensed too restrictively or when patents are used excessively to protect information "this could hamper research and development, clinical access, and availability of high-quality tests

[49] In accordance with these rules, the EPO considered that isolated DNA claims in Myriad's European BRCA patents were eligible. See The *University of Utah Research Foundation v. Institut Curie* (Mutation), 2008, T 0666/05 (EPO Board of Appeal).

[50] See C. LONG, *Patents and Cumulative Innovation*, in *Washington University Journal of Law and Policy,* 2, 2000, 229, 230-31.

[51] See also D.L. BURK, M.A. LEMLEY, *The Patent Crisis and How the Courts Can Solve It,* in *University of Chicago Press*, 2009, 73-75 (discussing cumulative innovation in patent law).

[52] See L. LESSIG, *The Architecture of Access to Scientific Knowledge, Lecture at Cern*, Geneve, Switzerland, 18 April 2001, available at http://www.youtube.com/watch?v=2me7hptVGzI (last visited 27/04/2021).

53See, e.g. *Bement v. Nat'l Harrow Co.*, 186 U.S. 70, 90-92 , 1902, ("The general rule is absolute freedom in the use or sale of rights under the patent laws of the United States").

[54] See L. BENTLY, B.SHERMAN, *Intellectual Property Law*, cit., 570. In the U.S., an intellectual property rights holder has no obligation to either use or license its property rights. On the point, see H. HOVENKAMP, M.D. JANIS, M.A. LEMELY, *Unilateral Refusals to License in the US*, in *Journal of Competition Law & Economics,* 2, 2006, 1, 13.

for patients".[55] The current Covid-19 global pandemic and lack of vaccine doses, is a clear example of the many challenges associated with exclusive rights, information sharing, affordability of medical treatment, and access to biotech innovations. This provide another piece of empirical evidence for the proposition that patents are not always an efficient means of protecting medical treatments having also a chilling effect on research and innovation imposing substantial barriers on other researchers' ability to undertake further investigation.[56] However, it is also true that in the absence of some form of protection or in the absence of other forms of flexibility, the only alternative would be to maintain secrecy with even more evident problems in this very restrictive scheme.[57]

5. The impact of court decisions for the Life Sciences Industry: Myriad and Mayo

As mentioned before, there are very few empirical studies on the real negative effects on gene-related patents resulting as a consequence of subject matter eligibility disputes.

Did Myriad really result in a reduction of gene-related patents in general (i.e. beyond isolated gene patents)? At first sight, the answer to this question seems to be negative. In particular, according to a study which has been published in 2016 on Nature Biotechnology, gene-related patents increased in number during the three-year period following the Myriad ruling.[58] In order to evaluate the impact and effect of the US supreme court decision, this study used an automated search algorithm intended to explore Myriad's impact by considering the record of granted gene-related patents using relevant key search terms before and after the decision. In addition, another study observed how over the 50% of gene-related patents granted in 2005 are already in the public domain demonstrating how patent protection may not be so important for development of genetic test.[59] This figure depicts that gene-related patent holders "often do not pay maintenance fees, meaning that their rights lapse and become part of the public domain before the full twenty years of patent life is realized".[60]

Another important aspect concerns the potential negative aspects deriving from the Myriad ruling on gene-related patents. In particular, it is interesting to analyse the trends in isolated gene patent activity (granted patents with gene-related claims) in the last 20 years in order to compare it with the trend after the decision. According to the data used by this study, from 2005 to 2016 the trend was

[55] See G. VAN OVERWALLE, *Turning Patent Swords into Shares*, in *Science,* 330, 2010, 1630.

[56] See D.B. RESNIK, *Owning the Genome: A Moral Analysis of DNA Patenting*, 2004, 141.

[57] See D.S. LEVINE, *Trade secrets and the battle against Covid*, in *Journal of Intellectual Property Law & Practice,* 15, 2020, 849 (proposing the creation of voluntary trade secret information sharing and/or compulsory trade secret licensing in order to face the unprecedented public health crisis due to Covid-19 pandemic).

[58] See M. ABOY et al., *Myriad's impact on gene patents*, in *Nature Biotechnology*, 34, 2016, 1119, 1120 (noting that the effects of the ruling on the biotech industry "have been less profound than some practitioners, scholars and patent holders anticipated"). An extended version of the study has been recently released, see J. LIDDICOAT et al., *The Effects of Myriad and Mayo on Molecular-Test Development in the United States and Europe: Interviews from the Frontline*, in *Vanderbilt Journal of Entertainment and Technology Law,* 22, 2020, 785 (analyzing how these court rulings affect business decisions surrounding the development of diagnostic tests).

[59] See J. LIDDICOAT et al., *The Effects of Myriad and Mayo on Molecular-Test Development in the United States and Europe: Interviews from the Frontline*, cit., 800.

[60] *Ibidem.*

down,[61] it means that the situation was not better before the Myriad ruling. Another question answered by this empirical study is whether Myriad negatively affected biotech patents activity such as diagnostic and biomarker patents. Also in this case, it seems that the ruling has not negatively affected the sector: in particular, data demonstrates that general biomarker patents continue to be issued in increasing number.[62]

Essentially these patent data are also useful to understand if – in case of a completely different decision of the Supreme Court – we could have observed also a different trend. In particular, the data on patent filling and granted dates versus priority dates seems to indicate that even if Myriad had reached the opposite decision, the patent activity directed to isolated gene patents would be very similar.[63]

The empirical analysis also considered additional variables such as the claim level analysis:[64] in particular, the analysis addressed the variability of patent claims after Myriad considering also the question if it is easy to draft around Myriad. In general, the results of the study show that in the years since Myriad "there has been much less amending activity than some commentators expected".[65] In over 79.2% of the patents containing isolated DNA claims were abandoned or all the isolated DNA claims were cancelled.[66] At the same time, only 18.6% of patents containing isolated DNA claims were successfully amended.[67] Finally, the investigation claimed that only 21 instances of successful amendments were found after receiving an explicit Myriad-based rejection and "none of these retained the scope (breadth) of the original applications".[68]

Another recent empirical investigation considered the impact of the Mayo ruling on patent prosecution with a particular attention to the future of patentability of diagnostic methods and personalized medicine treatments.[69] Although most legal scholars have described this decision as the tombstone for precision medicine,[70] there is little empirical evidence on these negative claims. The decision in Mayo has in fact opened many practical questions related to the future of biotech patents leaving patent applicants unsure of how to obtain and protect their intellectual property rights. According to the results of one of the few and structured empirical investigations based on a combination of keyword-based and semantic search, since July 2014 US patent examiner cited Mayo

[61] See M. ABOY et al., *Myriad's impact on gene patents*, cit., 1121.
[62] Ibidem.
[63] See M. ABOY et al., *Myriad's impact on gene patents*, cit., 1122.
[64] M. ABOY et al., *After Myriad, What Makes a Gene Patent Claim 'markedly different' from Nature,* cit., 822
[65] See for all, M. M. HOPKINS, et al., *DNA patenting: the end of an era?,* in *Nature Biotechnology*, 25, 2007, 185.
[66] See M. ABOY et al., *After Myriad, What Makes a Gene Patent Claim 'markedly different' from Nature*, cit., 822
[67] *Ibidem*.
[68] *Ibidem*.
[69] See MATEO ABOY et al., *Mayo's Impact on Patent Applications Related to Biotechnology, Diagnostics and Personalized Medicine*, in *Nature Biotechnology*, 37, 2019, 513.
[70] See e.g. E.J. HAANES, J.M. CANAVES, *Stealing Fire: A Retrospective Survey of Biotech Patent Claims in the Wake of Mayo v. Prometheus*, in *Nature Biotechnology*, 30, 2012, 758; J. L. FOX, *Industry Reels as Prometheus Falls and Myriad Faces Further Reviews*, in *Nature Biotechnology*, 30, 2012, 373; R.S. EISENBERG, *Diagnostics Need Not Apply*, in *Boston University Journal of Science & Technology Law*, 21, 2015, 256; C.M. HOLMAN, *The Mayo Framework Is Bad for Your Health*, in *George Mason Law Review*, 23, 2016, 901.

more than 33.000 times to justify rejection under 35 USC §101.[71] In addition, more than 38.000 patents document cited Mayo.[72]

Also in the case of the Mayo ruling – in order to evaluate the implications on the patent-eligibility matters – it was relevant to assess the real impact before and after the decision. According to the empirical results of this study, it seems there is an increase (from 10.5% to 55.5%) in the prevalence of 35 USC §101 rejections in key art units.[73] In addition, the analysis of patented, abandoned and pending patents (post-Mayo) reveals that 49.3% of the patent applications were rejected or abandoned; 27.6% were granted after a successful appeal to the examiner's Mayo-based rejection and 23.1% are still in active examination or prosecution.[74] These data seem to demonstrate that the Mayo decision has had a more significant impact than Myriad on patent prosecution in the life science sector. However, a substantial number of patent applications have been able to overcome objections raised by the Patent Office Examiners through amendments. This demonstrate that "it is possible to draft claim language that satisfies the post-Mayo 35 USC §101 threshold for life science inventions".[75]

Essentially, both these studies prove that – even if there are still many unresolved legal questions which contribute to maintaining high levels of uncertainty – the effects of the Myriad and Mayo decisions on gene patenting have been less extreme (at least at the macro-level) than many of the initial predictions.[76] Certainly, further empirical evidence is needed to fully explore the challenges posed by these decisions and possibly consider the necessity of a law reform. We can, however, say that these data also provide sufficiently convincing evidence to suggest that – at least in the case of genomic sequences – patents are not necessarily needed to foster the process of discovery of genomic elements. At the same time, it can be assumed that there is adequate space for patent protection of medical innovations without claiming genomic sequences.

6. The impact of court decisions for the Life Sciences Industry: Brüstle

Contrary to the cases decided by the US Supreme Court, the European controversies regarding the barriers to the patenting of stem cell research, seem to belong to the so-called phenomenon of the ethicalization of law. In fact, here it could be argued that the CJEU wanted to discourage medical research which requires the destruction of human embryos on the grounds that it is ethically unacceptable. The court basically held that any human embryonic stem cells invention which is not

[71] See M. ABOY et al., *Mayo's Impact on Patent Applications Related to Biotechnology, Diagnostics and Personalized Medicine*, in *Nature Biotechnology*, 37, 2019, 513, 514.

[72] *Ibidem*.

[73] *Ibidem*, 515.

[74] *Ibidem*.

[75] *Ibidem,* 516.

[76] Among the various voices suggesting a very negative impact, see e.g. M.M. HOPKINS et al., *DNA Patenting: The End of an Era?*, in *Nature Biotechnology*, 25, 2007, 185; I. HUYS et al., *Legal Uncertainty in the Area of Genetic Diagnostic Testing*, in *Nature Biotechnology*, 27, 2009, 903; J.L FOX, *Industry reels as Prometheus falls and Myriad faces further reviews*, in *Nature Biotechnology*, 30, 2012, 373; R.S. EISENBERG, *Diagnostics Need Not Apply*, in *Journal of Science and Technology,* 21, 2015, 256.

based on non-destructive techniques is not patentable,[77] so the ban on the patenting aims at pushing pharmaceutical research towards more (morally) acceptable practices.[78]

On the other hand, however, the stem cell biotech sector and other parts of the ecosystem responded negatively to the decision in *Brüstle* claiming that it would be able to jeopardise, and thus harm, research and development in a medical field which is considered fundamental and promising for the advancement of bio-medical knowledge.[79] The thing that everybody was worried about immediately after the ruling was a sort of brain drain from the EU towards more science and business friendly countries such as US, where there are no statutory limits on patent eligibility of human embryonic stem cells on moral or ordre public grounds.

Another criticism of *Brüstle* is based on an apparent conflict of fundamental rights. Human dignity is not the only fundamental right at stake here. Patents and in general intellectual property rights are also protected as fundamental rights, as confirmed by Article 17(2) of the EU Charter as well as Article 1 Protocol 1 of the European Convention of Human Rights (ECHR).[80] It has been argued that *Brüstle* has strongly limited the fundamental right to patents in the human embryonic stem cells field, and that a similar limitation should have been subject to a proportionality test similar to the one required by the EU Charter and the European Convention on Human Rights.[81] This test requires that any limitations of the fundamental right to properties (including patents) must not have a disproportionate impact on the owner in relation to the target sought. Yet, the CJEU in *Brüstle* made no reference to any proportionality test nor to any balancing exercise between the two rights in question.[82]

At present, it remains also uncertain whether concerns regarding commodification and dignity can be limited to stem cells or can be used in other cases including commercial uses of other human tissue products. Furthermore, the concept of human dignity reveals itself as equally complicated and multifaceted in particular when applied to the field of bioethics and human genetics.[83]

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[77] See S.H.E. HARMON et al., *Dignity, plurality and patentability: the unfinished story of Brüstle v Greenpeace*, in *European Law Review*, 38, 2013, 92, 99.

[78] See also A. PLOMER, *The European Union's IP Policy and Funding of Stem Cell Research*, in D. MATTHEWS, H. ZECH (eds.), *Research Handbook on Intellectual Property and the Life Sciences*, 2017, 230.

[79] See e.g. S. PARKER, P. ENGLAND, *Reaction to the Brüstle decision*, in *Pharmaceutical Patent Analyst,* 1, 2012, 233; T. FALTUS, *No patent-no therapy: a matter of moral and legal consistency within the European Union regarding the use of human embryonic stem cells*, in *Stem Cells and Development*, 23, 2014, 56.

[80] See Article 1 of the European Convention for the Protection of Human Rights and Fundamental Freedoms, as amended by Protocol 1, Sept. 3, 1953, E.T.S. 155 (EC). See also the case *Anheuser Bush v. Portugal* (2007), 45 E.H.R.R. 830, at para. 72 (E.C.H.R.) where the ECtHR held that "Article 1 of protocol No. 1 is applicable to intellectual property as such".

[81] See A. PLOMER, *After Brustle: EU Accession to the ECHR and the future of European patent law*, in *Queen Mary Journal of Intellectual Property*, 2, 2012, 110, 134.

[82] *Idem*. See also E. BONADIO, *Biotech Patents and Morality after Brüstle*, in *European Intellectual Property Review*, 34, 2012, 433.

[83] See J. MALPAS, N. LICKISS, *Introduction to a Conversation, in Perspectives on Human Dignity: A Conversation* 1 (J. MALPAS, N. LICKISS eds., 2007). For more on this topic, see e.g. C. FOSTER, *Human Dignity in Bioethics and Law*, Hart, 2011; M. NOGUEIRA DE BRITO, *Human Reproduction and Human Dignity as a Constitutional Concept,* in M.V. DE AZEVEDO CUNHA et. al. (eds.), *New Technologies and Human Rights*, 2013, 169.

Finally – although European patent law is relatively uniform – some aspects of patent eligibility of human embryonic stem cells seems to remain still inconsistent.[84] In particular – as has been observed by other scholars[85] – a closer look reveals that national divergences still prevail in the interpretation of the most recent case law on stem cells. As a result, new issues need to be solved at the various layers especially in relation to the institutional changes created by the unitary patent package – including the future role of the unified patent court in safeguarding coherence in the European patent system.[86]

It seems clear that the implementation of the moral exclusions will be somewhat problematic from a harmonization point of view, as the morality provisions will be applied by different decision-making bodies (Court of Justice of the European Union, Unified Patent Court, National courts) according different standards with possible conflicting interpretations.

7. Conclusion

Biotech patents raised, and still raise, delicate issues which have been analysed and commented by lawyers as well as legal and medical scholars. Such issues have been debated during patent procedures and disputes in Europe and US. Focuses and approaches in these countries have however been different and centered on distinct legal questions.

Intellectual property rights play an enormous role in the creation of diagnostics and treatments as well as in vaccine development. International organizations, member states and institutions must recognize that now is the time for a broader discussion on how to guarantee that sufficient quantities of these health products are produced, that they are affordable, and that they are equitably distributed globally.

It is probably time of framing this debate in a completely different manner. We are in fact dealing with a more prominent issue dealing on whether genes occupy a more central legal position. A need for a change of paradigm is also clearly necessary because precision medicine is changing the current model of drug development that is essentially based on a small number of very successful drugs that can be sold to millions of patients. As a consequence, lawmakers must realize that it is time to design a new and more efficient legal scheme able to effectively support the developing of these precision treatments.

As a matter of fact, the decisions of the US Supreme Court on biotech patents resulted in an overall lack of harmonization between Europe and the United States creating an asymmetry of subject-matter-eligibility regarding isolated gene patents. Under the new constitutional interpretation of the US isolated gene patents are no longer allowed whereas in European countries – according to the EU

[84] See U. STORZ, T. FALTUS, *Patent eligibility of stem cells in Europe: where do we stand after 8 years of case law?,* in *Regenerative Medicine*, 12, 2017, 37.

[85] See e.g. A. MCMAHON*, An institutional examination of the implications of the unitary patent package for the morality provisions: a fragmented future too far?*, in *International Review of Intellectual Property and Competition Law*, 48, 2017, 42.

[86] *Ibidem*.

Biotech Directive and the European Patent Convention – it is still substantially possible.[87] As noted by other authors, the issues raised in the US cases Myriad and Mayo could also be the occasion for "relight the fire surrounding the Biotech Directive".[88] In particular, as the COVID-19 pandemic is unfortunately showing us, the "product of nature" exclusion under patent law – which prevents the patenting of genomic sequence data – should be preserved and strengthened and possibly extended to other jurisdictions.[89]

At the same time, the US law has no equivalent ordre public or morality statutory provision capable of creating scientific or ethical roadblocks to human embryonic stem cell.[90] It means that U.S. does not currently have a formal ban on the use of germline editing techniques on human tissue while the European Biotechnology Directive deems technologies that require the destruction of a human embryo unpatentable. From this point of view, it is definitely strange that the country which at present has greatest doubts about biotech patent is also the one which does not take into account any ethical or deontological factors related to human embryonic stem cell patenting.

[87] Case T 1213/05, *Univ. of Utah Research Found. v. Sozialdemokratische Partei der Schweiz,* (EPO Boards of Appeal, 2007); Case T 0666/05, *The University of Utah Research Foundation v. Institut Curie* (Mutation) (EPO Board of Appeal 2008).

[88] See e.g. J.C. LAI, *Myriad Genetics and the BRCA Patents in Europe: The Implications of the U.S. Supreme Court Decision*, in *UC Irvine Law Review,* 5, 2015, 1041.

[89] See J. CONTRERAS, *COVID-19 as an Example of Why Genomic Sequence Data Should Remain Patent Ineligible*, in S. BURRIS et. al (eds.), *COVID-19 Policy Playbook: Legal Recommendations for a Safer, More Equitable Future, Public Health Law Watch*., 2020, 137.

[90] See Sherley & Deisher v. Sebelius, 644 F.3d. 388, 388-90 (D.C. Cir. 2011); Sherley & Deisher v. Sebelius, 689 F.3d. 776 (D.C. Cir 2012).

Special issue

From "familial searching" to "forensic genetic genealogy": New frontiers – and challenges – of DNA analysis in criminal investigations

Giulia Formici *

ABSTRACT: Since its discovery, DNA analysis has been an important tool in criminal investigations: the creation of national DNA databases, retaining the genetic profile of criminals, revealed to be crucial in solving serious crimes. In recent years, the expansive use of DNA analysis, together with scientific and technological progress, led to the development of new sophisticated investigative techniques, from the so-called "familial searching" to the more recent use of "forensic genetic genealogy", based on the exploitation of commercial genealogy databases by law enforcement authorities. Notwithstanding their effectiveness, these new instruments raise serious ethical and legal concerns: this paper aims at presenting these complex challenges, by underlying the need to strike a proper balance between the public interest to a rapid and efficient identification of unknown offenders and the dangerous shift towards "genetic surveillance".

KEYWORDS: DNA; genetic forensics; genetic genealogy databases; familial searching

SUMMARY: 1. DNA analysis and genetic databases: some preliminary information on the origins and functioning of a powerful crime-fighting tool – 2. The expansive use of DNA analysis in criminal investigations: ethical and legal implications of the *familial searching* technique – 2.1. Looking for "closeness" by endangering privacy, data protection, presumption of innocence and non-discrimination – 2.2. First efforts to enlighten the "shadow database" – 3. The controversial use of recreational genealogy databases in the US: emerging concerns – 3.1. The potentialities of *forensic genetic genealogy*: the *Golden State Killer* case – 3.2. *Forensic genetic genealogy* as an "outgrowth" of the *familial searching* technique: a way to step over safeguards regulating traditional DNA analysis? – 3.3. The Privacy Policies established by commercial genealogy companies and the limits of the "informed consent" – 4. How to avoid "genetic surveillance": paving the path towards a profound guarantee of "genetic privacy" – 4.1. The risks of a "universal database": some timid attempts of regulatory answers – 4.2. How to resist the temptation of "seeing into the life of citizens": prompting a thoughtful and pondered debate.

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The article was peer-reviewed by the editorial committee.

ISSN 2284-4503

1. DNA analysis and genetic databases: some preliminary information on the origins and functioning of a powerful crime-fighting tool

Since its first use in 1985,[1] DNA analysis has become a fundamental tool in criminal investigations. The so-called forensic genetics, based on the "ability to extract DNA profiles – a biological structure considered unique for every individual – from samples collected at crime scenes",[2] is of paramount importance to establish the unique "genetic identity" of an unknown perpetrator.

In order to expand the potentialities of forensic genetics, in the mid-1990s many Countries started to create national DNA databases retaining the genetic profiles of convicted persons and, in some States – depending on the different legislative solutions adopted –, also of arrestees, victims, missing people or persons of interests. These repositories can be accessed, at certain specific conditions, during criminal investigations for the purpose of comparing, through a mainly computerized process, the DNA sample and profile obtained from the crime scene or the victim's body to the genetic information stored in the national database, thus making it possible to identify the unknown offender in case of an exact positive match.[3]

Due to the very sensitive nature of genetic data, specific rules were approved by national legislators, disciplining the collection, retention ("eligibility criteria") and circulation of DNA profiles in national repositories and clearly establishing the conditions, procedural requirements and purposes which legitimize DNA databases to be accessed and used by specifically identified law enforcement

[1] In 1985, the Leicestershire police employed for the first time the DNA analysis to solve a violent crime (the rape and murder of two young girls). The "fathers" of the "genetic profiling" are Sir Alec Jeffreys, who discovered the first "genetic fingerprint" in 1984, and Peter Gill, who revealed the possibility to compare the DNA profile, developed from biological materials found on the crime scene, to a reference sample and profile belonging to a known person. For more information on the origins of DNA profiling and its use for criminal investigations, see, *ex multis*, G. CLARK, *Justice and science: trials and triumphs of DNA evidence*, New Brunswick, 2007; in Italian: L. SCAFFARDI, *Giustizia genetica e tutela della persona. Uno studio comparato sull"uso (e abuso) delle Banche dati del DNA a fini giudiziari*, Milano, 2017.

[2] H. MACHADO, R. GRANJA, *Forensic genetics in the governance of crime*, Singapore-Braga, 2020, 2. The authors clearly explain that "studies on the use of DNA for individual identification depends upon broad zones that exists between the genes that are generally called "non-coding DNA". These intergenic zones reveal specific chemical sequences that are supposed to be unique to each individual and therefore produce a "genetic fingerprint". Comparison of different genetic fingerprints enables us to observe whether different samples of DNA come from the same individual or different individuals. In short, each person"s DNA is unique, except in the case of identical twins", 46.

[3] The first forensic database was created in England, in 1995; according to a 2019 Interpol analysis, 70 Countries around the world have nowadays a national DNA database in place (INTERPOL, *Global DNA profiling survey*, 2019, https://bit.ly/39NpDPb). DNA databases usually contain only DNA profiles, whereas the biological samples and personal information related to the genetic profile are stored in a separate software or repository, for privacy and data protection reasons. For a broader analysis on the DNA databases" history and functioning, see P. MARTIN, H. SCHMITTER, P. SCHNEIDER, *A brief history of the formation of DNA databases in forensic science within Europe*, in *Forensic Science International*, 119, 2001, 225-231; M. HIBBERT, *DNA databanks: law enforcement"s greatest surveillance tool?*, in *Wake Forest Law Review*, 34, 1999, 767 ff.; N. VAN CAMP, K. DIERICKX, *National forensic databases: social-ethical challenges and current practices in the EU*, in *European Ethical-Legal Papers*, 9, 2007; R. HINDMARSH, B. PRAINSACK (eds), *Genetic suspects: global governance of forensic DNA profiling and databasing*, Cambridge, 2010.

authorities.[4] These complex rules and limits have usually been highly debated and discussed, because of their ability to affect, on the one hand, citizens' rights to privacy, data protection, presumption of innocence and non-discrimination, and, on the other hand, the efficacy of DNA forensic techniques: the wider the eligibility criteria are, the bigger the repository will be. If it is correct to affirm that the databases' size has an obvious positive impact on the possibility to find an exact match, it is also worth underlining that the retention of a vast amount of genetic information exposes sensitive data to significant risks of abuses, from data breaches to function creep. Finding a proper balance between the efficiency of this investigative instrument and a proportionate and necessary intrusion into citizens' private lives has represented a serious challenge for legislators, asked to take delicate regulatory decisions, sometimes challenged before national and supranational Courts. For example, in the European context, a relevant role has been played by the ECtHR: in the landmark case *S. and Marper v. UK,*[5] the Strasbourg Judges declared the UK legislation regulating the functioning of the *National DNA Database* (NDNAD) in violation of Art. 8 ECHR in so far as it authorized a blanket, indiscriminate and indefinite retention of DNA profiles belonging to merely suspected individuals, not subsequently convicted of offences.

This proportionality assessment and the correct balance between competing private and public interests led to different legislative solutions across the European Continent. In general terms, comparative surveys and analysis underlined the presence of two main tendencies: the so-called "extensive" and "restrictive" legislations,[6] able to impact on both the dimension of the national DNA database and the percentage of the population whose genetic profile can be included in the repository.[7]

[4] Together with technical standards (the quality and characteristics of data inserted in the database) and the conditions DNA laboratories should respect in order to be allowed to process genetic data.

[5] ECtHR Grand Chamber, 4 December 2008, Applications n. 30562/04 and 30566/04. According to the UK PACE (*Police and Criminal Evidence Act 1984*), genetic samples and DNA profiles of originally suspected but then unconvicted people could have been retained with no time limit and irrespective of the nature and gravity of the offence. On this decision: C. NYDICK, *The British invasion of privacy: DNA databases in the UK and in the USA in the wake of the Marper Case*, in *Emory International Law Review*, 23, 2010, 609-650; C. MCCARTNEY, *Of weighty reasons and indiscriminate blankets: the retention of DNA for forensic purposes*, in *The Howard Journal of Criminal Justice*, 51, 2012, 245-260.

[6] For a vast comparative analysis of national DNA databases" characteristics, with a focus on the European Continent, see: R. BROWNSWORD, *Genetic databases: one for all and all for one?*, in *King"s Law Journal*, 18, 2007, 273 ff; S. WALSH, J. BUCKLETON, O. RIBAUX, C. ROUX, T. RAYMOND, *Comparing the growth and effectiveness of forensic DNA databases*, in *Forensic Science International: Genetics*, 1, 2008, 667-678; F. SANTOS, H. MACHADO, S. SILVA, *Forensic DNA databases in European Countries: is size linked to performance?*, in *Life Sciences, Society and Policy*, 12, 2013, 1-13; ENFSI, *Survey on DNA-databases in Europe*, 2016; L. SCAFFARDI, *Giustizia genetica e tutela della persona*, cit.

[7] The elements that should be considered in order to determine the expansive or restrictive nature of national legislation governing DNA databases" eligibility criteria are related to: the criteria regulating the deletion of DNA profiles (f.i. after the death of the convicted or after a specific amount of years from the end of the conviction); the possibility to retain DNA profiles of merely suspected people or arrestees; the conditions for collection and retention of DNA profiles of people convicted (for all crimes or only for certain types of violent crimes, considered particularly serious); the deletion criteria of biological samples from which the DNA profile has been extracted; the scope of access; authorities allowed to access DNA databases.

In the US, the discipline of national DNA databases is far more complex: the *Combined DNA Index System* (CODIS) – the "FBI's program of support for criminal justice DNA databases as well as the software used to run these databases" – [8] is composed of a plurality of *DNA Index Systems*, hierarchically organized (national, state and local).[9] Along with federal legislation, establishing the eligibility criteria for the collection and retention of DNA profiles in the *National DNA Index System* (NDIS), there are also 50 DNA-collection State laws, creating a very fragmented regulatory landscape. As regards the federal discipline, the NDIS expanded its dimension after the approval of the *DNA Fingerprint Act* of 2005, which authorized any federal agency to collect DNA samples – and subsequently retain DNA profiles – not only from already convicted citizens (as recognized by the *DNA Identification Act* of 1994), but also from people arrested or facing charges for federal crimes. At the States' level, different practices can be registered: although all States have laws obliging convicted people to provide a DNA sample and include the DNA profile in the CODIS as well as in States' databases, some States' legislations limit this obligation only for certain convictions (usually violent crimes such as sex offences).[10] A broad inhomogeneity concerns the arrestees' discipline, with profound differences regarding the type of offences for which samples can be collected,[11] the possibility to store juveniles' profiles, and the rules – and procedural requirements – governing the deletion of DNA samples and the expungement of DNA profiles from genetic databases in case an arrest doesn't result in a final conviction.[12]

[8] See the *FBI fact sheets*, available at https://bit.ly/3uqwPc2.

[9] For an in-depth analysis of *Local DNA Index Systems* and the flow of genetic data from this lower level to *State DNA Index Systems* (SDISs) and the NDIS, see K. WAH, *A new investigative lead: familial searching as an effective crime-fighting tool*, in *Whittier Law Review*, 29, 2008, 909-960.

[10] "Forty-eight States require the collection of DNA for any felony conviction, and forty-two States require the collection of samples for at least some misdemeanor convictions", A. NIETO, *Familial searching: how implementing minimum safeguards ensures constitutionally-permissible use of this powerful investigative tool*, in *Cardozo Law Review*, 40, 2019, 1768.

[11] According to a study of the NCSL (National Conference of State Legislatures), updated to 2018 (available at https://www.ncsl.org/Documents/cj/ArresteeDNALaws.pdf), 31 States authorize, under specific conditions, the collection of DNA samples from arrestees. It"s important to mention that some States require *probable cause hearings* to establish the existence of a probable cause able to justify the DNA samples" collection. In this regard, the US Supreme Court was asked to evaluate the legitimacy of the State of Maryland law, imposing the collection of DNA samples from persons charged (but not already convicted) with burglary or violent crimes and requiring a judicial officer evaluation on the existence of a probable cause for the arrest. The Supreme Court, in *Maryland v. King* (2013), affirmed that such provisions don"t violate the Fourth Amendment and that a correct balance between public and private interests is established. As Joh clearly stated, this decision resulted in "opening up many opportunities for DNA collection by the police that extend beyond the limits of serious offences or even the category of arrestees", E. JOH, *Maryland v. King: policing and genetic privacy*, in *Ohio State Journal of Criminal Law*, 11, 2013, 294.

[12] The federal law imposes the expungement of DNA profiles from the NDIS once a conviction is overturned or, with reference to arrestees, the charge is dismissed or results in an acquittal. Similar provisions are established at the States" level: according to the abovementioned NCSL Survey, 16 States "provide for the expungement of a DNA record upon the request of the individual; 13 States provide for automatic expungement", https://www.ncsl.org/Documents/cj/ArresteeDNALaws.pdf. It is worth underlining that when automatic expungement is not in place, the arrestee has the burden of requiring expungement: nonetheless, people are usually unaware of this possibility or don"t have the necessary capabilities to start the request procedure.

Relying on the brief but necessary overview provided in this introductive Paragraph, the paper will focus on the challenges and issues posed by two most recent expansive evolutions of DNA analysis in criminal investigations: on the one hand, the use of genetic forensic beyond the determination of an exact match in a national DNA database,[13] implementing the so-called *familial searching* technique; on the other hand, the use of DNA analysis beyond national databases' searches, employing direct-to-consumer commercial genealogy databases with the purpose of detecting long-range familial relationships.

2. The expansive use of DNA analysis in criminal investigations: ethical and legal implications of the familial searching technique

2.1. Looking for "closeness" by endangering privacy, data protection, presumption of innocence and non-discrimination

Notwithstanding the efficacy of DNA analysis and the creation of national DNA databases, the traditional genetic forensic technique soon revealed its limits: when an exact match between the DNA profile collected from the crime scene or victims' body and the ones retained in the national repository is not found, no new and useful investigatory leads can be obtained and, in the absence of other evidence, the case risks to grow cold.

Starting from these premises and willing to exploit all the potentialities of genetic information, law enforcement authorities, together with geneticists, tried to develop innovative methods for testing and analysing genetic evidence. The so-called *familial searching* is one of the first sophisticated and controversial techniques deriving from an extensive use of DNA analysis and the adoption of different search parameters: unlike the traditional and routine searches in DNA repositories, looking for an exact match, this new investigative tool aims at detecting the likelihood of *genetic relatedness* through a low-stringency search. Based on the well-known genomics principle according to which we all share a significant portion of our genetic profile with other family members, this technique allows law enforcement agencies to find close relatives – siblings, parents and children – of the unknown offender by searching for a partial match[14] in the national DNA database, thus permitting to restrict the pool of potential suspects. In other words, *familial searching* represents an alternative and deliberate way of testing genetic information already stored in a criminal DNA repository,[15] different

[13] In the next Paragraphs, the general term "national criminal DNA database" will be used to refer to DNA databases run by public law enforcement authorities, mainly including convicted criminals, although, as previously seen, arrestees" or suspects" DNA profiles could also be included in these repositories, according to certain States laws.

[14] "A partial match suggests that the perpetrator of the crime and the offender in the database are related. [...] The greater the number of loci shared between two individuals, the greater the likelihood that the two individuals are related to one another", K. WAH, *A new investigative lead: familial searching as an effective crime-fighting tool*, cit., 922. For a very technical analysis of the technical functioning of *familial searching*, see also R. MATEEN, M. SABAR, S. HUSSAIN, R. PARVEEN, M. HUSSAIN, *Familial DNA analysis and criminal investigation: usage, downsides and privacy concerns*, in *Forensic Science International*, 318, 2021, 1-6.

[15] In other words, "In crime investigations, familial searching is defined as the intentional search of an offender DNA database for inexact matches between DNA evidence profiles and offender and arrestee DNA profiles. Upon the identification of one or more partial match profiles, law enforcement may investigate a purported

from a random partial matching.[16] The efficacy of this instrument depends upon different elements, among which the dimension, the eligibility criteria and the accuracy of national DNA databases: the bigger a national database is, the more the possibilities to find partial matches with already convicted criminals' profiles.

In recent years, a lot of newspapers reported an appreciable number of crimes solved using the *familial searching* technique,[17] mostly in the UK and US, and especially with reference to the so-called "cold cases"; in addition, law enforcement authorities underlined the fundamental impact of *familial searching* for investigations concerning missing people or for dismissing charges against wrongfully convicted people or suspects.

Despite these positive impacts, the technique has been profoundly debated by academics, legislators, Courts and civil society. As highlighted by many Civil Liberties Groups, *familial searching* implies the creation and exploitation of a sort of an unofficial "shadow DNA database",[18] made of (partial) genetic information pertaining to innocent people who simply are in close kinship connection to convicted criminals.[19] The use of this "shadow database" permits to multiply the potentialities of genetic testing by extending the number of possible matches related to a single genetic profile: in other words, the sentenced offender become a "genetic informant", unintentionally and indirectly targeting strict relatives and subjecting them to testing and investigations – and eventually implicating them –. In this sense, the *familial searching* materialises in "an expansion of the net of genetic surveillance to [mainly innocent] persons whose genetic information would have remained private from the State has it not been for the actions of their blood relatives".[20] By doing so, this technique, if routinely applied, can be employed to circumvent limits and safeguards already provided for the "traditional" use, collection, retention and access to genetic profiles and ultimately raises serious legal and ethical concerns related to a proportionate and necessary impact on the rights to privacy, data protection, but also on the presumption of innocence and non-discrimination.

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family member of the partial matches as suspects", J. KIM, D. MANNMO, M. SIEGEL, S. KATSANIS, *Policy implications for familial searching*, in *Investigative Genetics*, 2, 2011, 2.

[16] There is a relevant difference from the so-called "partial matching" technique, *per se* considered, and the *familial searching*: "partial matching frequently occurs by accident whether as recognized by an analyst or by a quality assurance measure; whereas familial searching is a deliberate database search for family members to generate investigative leads", R. WICKENHEISER, *Forensic genealogy, bioethics and the Golden State Killer case*, in *Forensic Science International: Synergy*, 1, 2019, 117.

[17] For some examples of famous crimes solved thanks to *familial searching*, see A. NIETO, *Familial searching: how implementing minimum safeguards ensures constitutionally-permissible use of this powerful investigative tool*, cit., and in K. WAH, *A new investigative lead: familial searching as an effective crime-fighting tool*, cit.

[18] This suggestive expression has been used by E. MURPHY, *Relative doubt: familial searches of DNA databases*, in *Michigan Law Review*, 109, 2010, 291-348 (see also, E. MURPHY, *The New Forensics: Criminal Justice, False Certainty, and the Second Generation of Scientific Evidence*, in *California Law Review*, 95, 2007, 1-71).

[19] The genetic information could also be referred to individuals merely suspected or arrested, depending on the rules governing the database"s eligibility criteria, as underlined in Paragraph 1.

[20] H. MACHADO, S. SILVA, *What influences public views on forensic DNA testing in the criminal field? A scoping review of quantitative evidence*, in *Human Genomics*, 13, 2019, 2.

If the legitimacy of a forced collection of DNA profiles from people convicted for a crime is highly undebated, considering that these persons have "forsaken their right to privacy",[21] the same cannot be said for other kinds of searches or genetic testing and analysis – such as the *familial searching* – that can lead towards the dangerous shift of a vast "genetic surveillance".[22] Moreover, this technique results having a stronger and more invasive impact on certain ethnic groups – such as black people and Hispanic –, due to the higher representation of minorities' genetic profiles in national DNA databases. Consequently, *familial searching* has been considered a dangerous investigative instrument, able to exacerbate, especially in the US context, already existent "disparities in the criminal justice system, in which arrests and convictions differ widely based on race, ethnicity, geographic location and social class".[23]

On the basis of these possible side effects and despite the potentialities of this instrument, able to invaluably support law enforcement authorities when all other possible investigative leads fail to identify a suspect, the opportunity to apply *familial searching* should be seriously and carefully evaluated, by also taking into proper account both the risks of false identification[24] and the elevate costs in terms of money, time and human resources required to assess and select that information really and concretely useful and relevant, among the vast range of results possibly produced.

2.2. First efforts to enlighten the "shadow databases"

Familial searching was first used as a crime investigation tool in UK in 2002 and immediately raised the attention of bioethicists, lawyers and civil society: the Nuffield Council,[25] in its Report "The forensic use of bioinformation: ethical issues", clearly highlighted doubts and perils deriving from the employment of the *familial searching* technique, urging for dedicated policies, a strong ethical oversight and detailed and independent researches both on its concrete usefulness and on the

[21] D. SYNDERCOMBE COURT, *Forensic genealogy: some serious concerns*, in *Forensic Science International: Genetics*, 36, 2018, 203. According to an affirmed doctrine and case-law, in the US "pivot persons, who have been convicted of one of the classes of crimes under which a DNA sample can be compelled, have a reduced expectation of privacy that is substantially outweighed by society"s interest in identifying the offender [...] Two US Supreme Court cases have recognized that the rights of these individuals are diminished to the extent that their rights are fundamentally inconsistent with the needs and exigencies of the regime to which they have been lawfully committed", K. WAH, *A new investigative lead: familial searching as an effective crime-fighting tool*, cit., 937.

[22] This expression has been used by J. ROSEN, *Genetic surveillance for all*, in *Slate*, 17 March 2009.

[23] F. BIEBER, C. BRENNER, D. LAZER, *Finding criminals through DNA of their relatives*, in *Science*, 312, 2006. On this topic, see also D. GRIMM, *The demographics of genetic surveillance: familial DNA testing and the Hispanic community*, in *Columbia Law Review*, 5, 2007, 1164-1194.

[24] As Syndercombe Court reported, "a false Y chromosome match in the case of Chen Long-Qui, for example, led to him being wrongly imprisoned in Taiwan for four years. [..] Using this approach to uncover relatives may not be that simple if the relationship is more distant", underlining the technical limits and the intrinsic possible error rate that characterise this instrument (D. SYNDERCOMBE COURT, *Forensic genealogy: some serious concerns*, cit., 203).

[25] The Nuffield Council on Bioethics is an independent body that evaluates and prepares studies on sensitive ethical issues related to biology, medicine as well as to the use of biometric and genetic data for scientific research or investigative purposes.

practical consequences for fundamental rights.[26] For these reasons, the Association of Chief Police Officers, the Home Office, the Information Commissioner and representatives from the Human Genetics Commission established specific rules and conditions upon which familial searches are allowed: although the details of this policy are not completely publicly available, surveys and consultations revealed that this controversial investigative technique should be approved – not automatically but on a case-by-case basis – by the UK Chairman of the Database Strategy Board,[27] only for serious crimes and only once all other possible – less invasive – instruments failed to reveal useful investigative leads.[28] The prior authorization procedure, aiming at avoiding the transformation of *familial searching* into a routine assessment, rely on the notions of "seriousness of crime" and "sufficient resources": the Chairman is required to assess not only the level of gravity of the specific case but also the concrete capabilities of law enforcement authorities – in terms of human resources, time and money – to fully develop useful investigative leads from the – usually complex and multiple results of the familial searches.[29]

[26] The Council didn"t recommend a total ban of this instrument, but suggested that this technique "is not used unless it is necessary and proportionate in a particular case", NUFFIELD COUNCIL, *The forensic use of bioinformation: ethical issues*, 2018, 79.

[27] The Strategy Board "provides governance and oversight over the operation of the NDNAD [...]. The Board comprises representatives of the National Police Chief"s Council, the Home Office, the DNA Ethics Group, the Association of Police and Crime Commissioners, the Forensic Science Regulation, the Information Commissioner"s Office, the Biometrics Commissioner, representatives from the police and devolved administrations of Scotland and Northern Ireland", in https://www.gov.uk/government/groups/national-dna-database-strategy-board.

[28] "In considering whether to approve the application, the Chairman will consider the nature and gravity of the crime and whether there is a need to explore every investigative avenue to identify the offender, as well as the availability of funding and resources to pursue the search", T. PIQUADO, C. MATTHIES, L. STRANG, S. ANDERSON, *Forensic familial and moderate stringency DNA searches. Policies and practices in the US, England and Wales*, Santa Monica, 2019.

[29] Since the precise conditions regulating the current policy on *familial searching* are not fully accessible, the public debate over this investigative instrument is only based on the few reports and surveys available: see for example C. MAGUIRE, L. MCCALLUM, J. WHITAKER, *Familial searching: a specialist forensic DNA profiling service utilising the NDNAD to identify unknown offenders via their relatives. The UK experience*, in *Forensic Science International: Genetics*, 8, 2014, 1-9; R. GRANJA, H. MACHADO, *Ethical controversies of familial searching: the views of stakeholders in the UK and in Poland*, in *Science, Technology and Human Values*, 6, 2019, 1068-1092; T. PIQUADO, C. MATTHIES, L. STRANG, S. ANDERSON, *Forensic familial and moderate stringency DNA searches. Policies and practices in the US, England and Wales*, cit. It should be noted that, according to an official report, "Since the technique was implemented in 2002, more than 200 investigations have been conducted, assisting in the resolution of about 40 criminal cases (data from 2012) in the UK", O. GARCIA, M. CRESPILLO, I. YURREBASO, *Suspects identification through "familial searching" in DNA databases of criminal interest. Social, ethical and scientific implications*, in *Revista Espanola de Medicina Legal*, 1, 2017, 26-34. Notwithstanding this relevant employment and the lack of transparency on the specific rules agreed by the different components of the UK Board, it is worth mentioning that the UK is one of the few European Countries having a specific *familial searching* policy. For the analysis of the disciplines adopted in other European States, see: O. GARCIA, M. CRESPILLO, I. YURREBASO, *Suspects identification through "familial searching" in DNA databases of criminal interest. Social, ethical and scientific implications*, cit.; H. MACHADO, R. GRANJA, *Forensic genetics in the governance of crime*, cit. and T. PIQUADO, C. MATTHIES, L. STRANG, S. ANDERSON, *Forensic familial and moderate stringency DNA searches. Policies and practices in the US, England and Wales*, cit.

In the US, this investigative method has often been subject to specific policies and, in certain cases, even to a general ban: at the federal level, for example, *familial searching* cannot be conducted, but the FBI left room for different disciplines adopted at the States' level, with the limit that only States' DNA databases can be subject to such searches (consequently excluding the genetic information contained in the NDIS). In this context, Maryland and Washington DC[30] are the only States which approved specific laws prohibiting the use of *familial searching*. On the contrary, and similarly to the UK discipline, in California the first specific policy regulating this tool was approved in 2008: although it is *per se* allowed, this technique is limited only to most serious crimes and should pass the prior administrative control of dedicated *State-Committee*, comprised of scientists, attorneys and law enforcement agents, whose task is to evaluate if the results produced by the *familial searching* can be actually useful and if these searches are able to effectively contribute to opening new investigative leads; moreover, it is established that incidental findings – such as non-paternity – are not disclosed to local law enforcement authorities, so as to exclude unnecessary intrusions into the private sphere of potential suspects.[31] Most recently, the New York State's law enforcement agencies adopted in 2017 a *Familial Searching Policy*, requiring the District Attorney to certify that all possible investigative efforts have been made in order to avoid *familial searching*, which should be considered as a "last resort" to be implemented when all other – possibly less intrusive – investigative methods resulted unsuccessful.[32] The fragmented approach that characterizes the US regulatory panorama, dominated by inhomogeneous policies' choices and different levels of safeguards and limits, brings to light the disadvantages associated with the absence of a federal law, able to provide shared guidelines and restrictions on such a delicate investigative tool.

Notwithstanding the different attempts to "enlighten", through the adoption of specific safeguards and conditions, the "shadow database", the debate on the limits and risks represented by *familial searching* is still widely open: while some commentators concluded that "familial searches should be forbidden because they embody the very presumptions that our constitutional and evidentiary rules have long endeavored to counteract: guilt by association, racial discrimination, propensity, and even biological determinism",[33] other scholars have considered it acceptable to employ closeness searches

[30] The Search Code of Maryland established, in 2010, that "A person may not perform a search of the statewide DNA data base for the purpose of identification of an offender in connection with a crime for which the offender may be a biological relative of the individual from whom the DNA sample was acquired", Public Safety Code § 2-506, lett. d). The 2012 Washington DC Code, § 22-4151, affirms that "DNA collected by an agency of the District of Columbia shall not be searched for the purpose of identifying a family member related to the individual from whom the DNA sample was acquired" (lett. b).

[31] E. MURPHY, *Law and policy oversight of familial searches in recreational genealogy databases*, in *Forensic Science International*, 292, 2018, e6.

[32] Other States performing *familial searching* are Arizona, Arkansas, Colorado, Florida, Michigan, Texas, Utah, Virginia, Wisconsin and Wyoming. For an in-depth analysis of policies and choices taken at the US States" level, see B. FIELD, S. SEERA, C. NGUYEN, S. DEBUS-SHERRIL, *Study of familial searching policies and practices: case study brief series*, ICF Paper, August 2017.

[33] E. MURPHY, *Relative doubt: familial searches of DNA databases*, cit., 34. The author has affirmed that "a Court might deem irrational a formal policy that effectively divides the population into two groups – those related to convicted offenders and those who are not – and then threats the former population as presumptive suspects in criminal investigations while exempting the latter population from such suspicion" (331). Murphy also criticized some of the policies and safeguards adopted in certain States to limit the impact of *familial searching*:

provided that stringent limits are determined, such as the proportionality of the intrusion evaluated on a case-by-case basis,[34] the seriousness of the crime[35] and the existence of proper procedural and privacy safeguards. Other commentators, on the contrary, rejected what have been defined as "myths and exaggerations"[36] regarding *familial searching*: this technique cannot be considered as equivalent to a "guilt by association" investigative method since it only represents an instrument to generate new leads and not a list of precise suspects. Moreover, according to Wah, relatives contacted by law enforcement authorities after a partial match has been found, "may decline to answer questions or leave",[37] so that any unreasonable search and seizure occur. Consequently, no constitutional issues derive from *familial searching*, *per se*: although a "humane system of criminal justice should strive to keep side effects to a minimum, consistent with the objective of convicting the guilty", "forgoing the opportunity to apprehend and prosecute wrongdoers also has grave costs. An advanced database system that includes highly accurate kinship matching is a permissible legislative choice", also considering that in almost all jurisdictions, specific rules preventing misuses of genetic data and ensuring a high-quality standard of laboratories and analysis are in place.[38] Finally, the same authors consider the discriminatory impact of the *familial searching* as a false affirmation: far from being related to this techniques, the over-representation of specific minorities in DNA databases is related to more profound and already existent problems affecting the criminal justice system; using DNA analysis such as the *familial searching* can, on the contrary, represent a

for example, "allowing executive branch officials - whether a governor, attorney general, or state laboratory administrator - to unilaterally authorize such a wide-sweeping and politically contentious form of searching is to grant the executive unchecked authority to dramatically expand the size and character of the DNA database", 341. Differently from other authors who considered the prior authorization a viable and useful safeguard, the idea of a mere administrative control by public authorities, such as the attorney general, is not considered a sufficient measure to avoid risks of abuses.

[34] See, among the others, S. SUTER, *All in the family: privacy and DNA familial searching*, in *Harvard Journal of Law and Technology*, 2, 2010, 310-399 and A. NIETO, *Familial searching: how implementing minimum safeguards ensures constitutionally-permissible use of this powerful investigative tool*, cit. The latter author, considering the choice of some US States not to allow *familial searching*, affirms that "if properly drafted and scrupulously monitored, familial searches policies have the potential to solve cold cases and exonerate individuals who have been wrongly convicted", thus expecting "law enforcement in States that have banned familial searches to urge lawmakers in their States to adopt such policies in the future", 1770. Both authors consider that, in the US context, *familial searching* would withstand a Fourth Amendment challenge.

[35] It is nonetheless worth underlining that some US States, such as Virginia, allow familial searches to be conducted against convicted criminals as well as arrestees. Other US States, for example Colorado, don"t limit *familial searching* to serious crimes "but requires that crime investigators submit written requests to conduct familial searching when the crime under investigation poses a substantial public safety concern and conventional investigative approaches have been exhausted", J. KIM, D. MANNMO, M. SIEGEL, S. KATSANIS, *Policy implications for familial searching*, cit., 6. The authors suggest some possible solutions and specific policies useful to increase efficiency without undermining fundamental rights, proposing some limitations – such as the creation of a specific ethical committee or a limited list of crimes legitimizing the use of *familial searching* – able to address legal and social concerns.

[36] D. KAYE, *The genealogy detectives: a constitutional analysis of familial searching*, in *American Criminal Law Review*, 50, 2013, 160.

[37] K. WAH, *A new investigative lead: familial searching as an effective crime-fighting tool*, cit., 941.

[38] D. KAYE, *The genealogy detectives: a constitutional analysis of familial searching*, cit., 163. The author firmly opposes Murphy"s theories and her definition of "shadow database".

means to alleviate these issues: "investigators will be guided by the DNA evidence, which holds no preconceived notions, stereotypes or biases about racial, ethnic or social groups and classes".[39]
This ongoing debate appears as the result of a dichotomy between public safety and efficiency of law enforcement activities, on the one hand, and fundamental rights' guarantees on the other hand, that urgently requires the assessment of a clear balance-point.

3. The controversial use of recreational genealogy databases in the US: emerging concerns

3.1. The potentialities of forensic genetic genealogy: the Golden State Killer case

In recent years, the ethical and legal debate arisen from the extensive use of DNA analysis through the *familial searching* technique has witnessed a significant and cumbersome development: the emergence of what has been significantly named *forensic genetic genealogy.*[40] This new investigative instrument exploits the great potentialities of genetic genealogy and, in particular, the unprecedented source of information represented by the commercial and recreational genealogical databases, run by private companies. Differently from traditional forensic DNA analysis, "the power of genetic genealogy lies in the comparison process and the ability to search for genetic matches in databases", by combining genealogical research techniques with the information contained in DNA profiles for the purpose of detecting existent biological relationships.[41]

[39] K. WAH, *A new investigative lead: familial searching as an effective crime-fighting tool*, cit. 955. Seemingly, Nieto stated that "it is an unfortunate reality that the American criminal justice system is heavily racialized [...]. Familial searches are a part of this imperfect system, and until a massive overhaul of the criminal justice system truly changes this reality, it is left to states and law enforcement to ensure that their actions and policies provide equal treatment to the greater extent possible", A. NIETO, *Familial searching: how implementing minimum safeguards ensures constitutionally-permissible use of this powerful investigative tool*, cit., 1790. For these reasons, the author affirms that *familial searching* cannot be, *per se*, considered to have a discriminatory purpose and the adoption of appropriate safeguards can ensure sufficient guarantees of equality in the implementation of *familial searching* techniques.

[40] See, for example, C. PHILLIPS, *The Golden State Killer investigation and the nascent field of forensic genealogy*, in *Forensic Science International: Genetics*, 36, 2018, 186-188; D. SYNDERCOMBE COURT, *Forensic genealogy: some serious concerns*, in *Forensic Science International: Genetics*, cit. Other authors use the term "forensic DNA phenotyping", referred to "a set of techniques that allow inferring genetic ancestry and externally visible characteristics of criminal suspects on the basis of a DNA sample", H. MACHADO, R. GRANJA, *Forensic genetics in the governance of crime*, cit., 86.

[41] For a deep yet understandable explanation of how DNA testing and genetic genealogy work, see D. KENNET, *Choosing your DNA test: the best DNA testing kits*, May 2020, https://bit.ly/3dFBjor. It is worth mentioning that "traditional genealogy has been practiced for centuries, using documentary records and oral histories to trace families backwards in time. Until recently, they were the only ways to connect extended family members, but with the advent of direct-to-consumer genetic testing, it is now possible to find relatives through shared DNA", E.M. GREYTAK, C. MOORE, S. L. ARMENTROUT, *Genetic genealogy for cold case and active investigations*, in *Forensic science international*, 299, 2019, 104. From a very technical point of view, unlike the traditional forensic DNA analysis technique which uses *autosomal short tandem repeats* (so-called STRs method) in order to determine a genetic profile, genetic genealogy employs a great amount of *single nucleotide polymorphisms* (SNPs method) disseminated in the autosome, able to offer a vast variety of information, from the identification of distant relatives to the prediction of pathologies" predisposition (such as Alzheimer or mental diseases). This difference is strictly related to the very nature and purpose of the two different analysis" methods: if traditional DNA profiling used by law enforcement authorities aims mainly at identifying offenders and intends

Essentially, consumers of commercial genealogical services are asked to submit and upload a genetic sample – in some cases via cheek swab or spit kit, usually provided by the company itself, or in other cases a sample generated from other sources – to genealogy companies, such as AncestryDNA, 23andMe, MyHeritage and FTDNA, in order to obtain a DNA testing; this preliminary analysis, based on high-quality DNA sample – substantially different from the ones usually found on the crime scene, often small and of degraded quality –, is functional to a subsequent and delicate phase, that of the comparison of consumer's markers to other users' profiles stored in the company database. This second operation "provides the user with a list of DNA matches and a prediction of the possible relationship or range relationships based on the amount of DNA shared".[42] The more extended a direct-to-consumer company repository is, the more matches can be found.[43] Although errors and misattributed relationships are a concrete possibility, these DNA databases have become increasingly used to find unknown relatives and to build a precise family tree (descendancy research), also thanks to more affordable DNA testing costs.[44]

The spread and growth of these databases have inevitably augmented the number of genealogical records available online; this tendency has also led to the creation of open-data genomics DNA databases, such as GEDmatch: unlike direct-to-consumer companies, these services don't provide for genetic testing; they only guarantee a comparison between a DNA genealogy test, uploaded by the user, and the genetic data voluntarily made available by other consumers who opted in to share their profiles and identities. The algorithms employed by the online database allow a one-to-many query and research, returning the user a list of other customers whose DNA information presents more matches with, also specifying the estimated relationship and/or the amount of DNA shared.

For their potentialities, related to the quality of genetic data retained (the so-called "density" of genetic marker data) and the broad dimensions of the genealogical databases, these repositories and the connected genetic genealogy techniques have recently drawn the law enforcement authorities' attention, especially as regards cold cases' investigations: once a perfect match with the DNA profile of the offender cannot be detected in national criminal DNA databases, and/or the *familial searching* method has not been effective – meaning that no relatives of the offender have been subject to a conviction in the past or their DNA profiles have not been included in the criminal genetic database, the access to commercial genealogical repositories, by possibly giving information about the

to limit its impact on the intimate sphere of citizens, using only the less invasive STRs method, in contrast "SNPs are chosen precisely for their informational richness. People submit their DNA to sites like 23andMe or MyHeritage because they want to know more about their genetic make-up than just identity. 23andMe, for instance, offers information about disease carrier status, predictive wellness, and cosmetic conditions, relying on hundreds of thousands of SNPs rather than the 13-20 STRs in the typical forensic profile", E. MURPHY, *Law and policy oversight of familial searches in recreational genealogy databases*, cit., e5.

[42] D. KENNET, *Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes*, in *Forensic Science International*, 301, 2019, 108.

[43] As underlined in the 23andMe website, "The 23andMe DNA database has more than five million genotyped customers worldwide. You will continue to find new relatives as our database grows over time", https://www.23andme.com/en-int/dna-ancestry/.

[44] "By February 2019 it was estimated that more than 26 million people had taken a direct-to-consumer genetic test. By 2021 there are likely to be over 100 million people in the direct-to-consumer databases", D. KENNET, *Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes*, cit., 108.

genealogy of the unknown offenders, can be a valuable tool, able to narrow the suspects' pool to a – or more – specific family tree.

It comes with no surprise that in recent years, genealogical databases have been used by US criminal investigators to detect new suspects' leads in numerous active cases or to "revitalize" cold cases – mostly related to missing people or serious crimes, some of which remained unsolved for decades – [45]. Among them, what has become famously known as the *Golden State Killer* case had the merit to shed light on this new investigative tool, by bringing the nascent technique to the public attention, stimulating a profound and paramount debate.[46]

In April 2018 the California police declared the arrest of Joseph James DeAngelo, accused to be the notorious serial killer and rapist of more than 50 women in California, from the 1970s to the 80s. Despite DNA samples – and consequently the profile – of the offender were found in multiple crime scenes, no match between the killer's DNA and the genetic profiles retained in the NDIS was found and no suspect was identified. For this reason, police investigators decided to upload the genetic profile of the offender on GEDMatch, by creating a false user account and identity:[47] the matching genealogical operations gave a considerable number of results, which were then employed by investigators, together with genealogists, to construct a potential offender's family tree and to gradually narrow, through traditional investigative techniques, a possible suspects list.[48] After months of complex researches, the police reached Joseph DeAngelo and, using a discarded DNA sample,[49] finally obtained a direct and exact match between the suspect's profile and the DNA found

[45] For some relevant case studies and examples of crimes solved thanks to the use of this particular technique, see D. KENNET, *Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes*, cit.; but also E.M. GREYTAK, C. MOORE, S. L. ARMENTROUT, *Genetic genealogy for cold case and active investigations*, cit., 110 ff.

[46] As underlined by Machado and Granja, "the case was considered by *Nature* one of the scientific events that shaped the year of 2018. Barbara Rae-Venter, a genealogist who helped to identify the golden State Killer, was distinguished by the same journal as one of the "ten people who mattered this year". According to *Time*, Barbara Rae-Venter "has provided law enforcement with its most revolutionary tool since the advent of forensic DNA testing in the 1980s"", H. MACHADO, R. GRANJA, *Forensic genetics in the governance of crime*, cit., 91.

[47] While uploading the unknown offender"s DNA profile, investigators had to declare, according to the Terms and Conditions of the direct-to-consumer service, that the genetic data were either a) their own; b) that they were the legal guardian of the DNA donor or c) that they were authorized for other reasons. None of these affirmations were true in the *Golden State Killer* case.

[48] As reported by Kennet and by many newspapers which disclosed important details related to the *Golden State Killer* case, "several thousand hours of genealogical detective work" were required in order to "build" a clear family tree, starting from very distant matches (D. KENNET, *Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes*, cit., 107); on this point, see also R. CARROL, *Golden State Killer: hope for unsolved murder case as ex-cop arrested*, in *The Guardian*, 26 April 2018; G. KOLATA, *The Golden State Killer is tracked through a thicket of DNA and experts shudder*, in *New York Times*, 27 April 2018; J. JOUVENAL, *To find alleged Golden State Killer, investigators first found his great great great grandparents*, in *The Washington Post*, 1 May 2018.

[49] In the US, during investigations and without a consent of the interested individual, it is considered lawful to employ "discarded" DNA, namely an abandoned biological sample (for example the DNA found on a cigarette-tip or on a glass or a chewing gum). This is motivated by the enforcement of the so-called "third-party doctrine", related to the right to privacy and the Fourth Amendment protection: the idea is that once a biological sample is discarded, a person cannot invoke the existence of a "reasonable expectation of privacy"

on the crime scene. After the arrest, in 2018, DeAngelo pleaded guilty to 13 counts of first degree murders in June 2020.

3.2. Forensic genetic genealogy as an "outgrowth" of the familial searching technique: a way to step over safeguards regulating traditional DNA analysis?

The great mediatic attention dedicated to this case, together with the profound debate it spurred on privacy, ethical, societal and legal concerns deriving from the employment of commercial third-party genealogical databases for investigative purposes, impose a serious analysis of the risks and perils connected to a possible future implementation and extensive use of this technique. Even if some of these issues are similar to the ones already underlined with regards to the *familial searching* instrument, the *forensic genetic genealogy* brings new and additional challenges, due to the peculiar nature, extension and data retained in the recreational databases searched by law enforcement authorities.

In this regard, *forensic genetic genealogy* can be considered an "outgrowth"[50] of familial searching of government-run criminal DNA databases but, differently from the latter, the former technique offers expanded potentialities – and dangers –: while a familial search in the NDIS repository can identify, at best, siblings, parents or children related to the offender's DNA profile, the application of genetic genealogy to commercial databases' information can trace thousands of relatives. As a result, "the sheer number of persons who must be investigated, and the amount of information law enforcement must amass on those persons in order to winnow down candidates, far exceeds that of a typical familial search".[51] Although these characteristics – able to detect a significant number of long-distance relatives starting from the unknown offender's genetic profile – represent the strength and the most relevant potential of the *forensic genetic genealogy,*[52] they also unveil the vast intrusiveness of this instrument. Unlike the *familial searching*, which results effective only if the

over his/her DNA. On this well affirmed, yet debated, theory, see more broadly E. JOH, *Reclaiming "abandoned" DNA: the Fourth Amendment and genetic privacy*, in *North Western University Law Review*, 2, 2006, 857-884.

[50] E. MURPHY, *Law and policy oversight of familial searches in recreational genealogy databases*, cit., e5.

[51] *Ivi*, e6; the author underlined that in the *Golden State Killer* case the investigators "mapped thousands of relatives, creating 25 distinct lines on the family tree"; in addition, during the investigations "at least two persons had their DNA sampled as a result of false leads in the database", and other genealogical databases have been searched (Ysearch for example) together with GEDmatch; these searches also led to an innocent 73-years-old man, living in Oregon, who was wrongfully identified and searched, being totally unrelated to the case. These exemplifications contribute to deeply understand the vast amount of individuals that can be subjected to investigations and searches thanks to the use of *forensic genetic genealogy*.

[52] From a technical point of view, some authors underlined that "even when the only matches are distant and large family trees must be constructed because common ancestors are many generations in the past, genetic genealogists can triangulate among the matches to determine the most promising branches of the family tree [...]. Even for perpetrators who are completely under the radar or long dead, given DNA from a crime scene, it may be possible to identify them with genetic genealogy [...]. Looking to the future, genetic genealogy has the potential to significantly reduce the number of unsolved cold cases in North America while also reducing the rate at which cases go cold", E. M. GREYTAK, C. MOORE, S. L. ARMENTROUT, *Genetic genealogy for cold case and active investigations*, cit., 113.

unknown offender has relatives already sentenced for a crime,[53] in the *forensic genetic genealogy* technique people whose DNA profile is compared to the unknown offender's one are individuals – not necessarily connected or related to a convicted person – who decided to upload their genetic information only for medical testing or ancestry detection. In addition, it should be considered that "individuals are often aware of even distant family members' arrests. This is especially true for violent or serious crimes, when police can take DNA at the time of arrest. By contrast, a person may not know that an immediate family member sent a DNA sample to a company for medical testing or ancestry analysis"[54] and consequently cannot know in advance to what extent his/her privacy is exposed because of a – even distant – relative's decision to upload the genetic profile in a private genealogy repository.

From all these considerations, it is clear that genetic genealogy as investigative tool entails a much more profound impact in terms of number of people involved, potentially subjecting thousands of innocent users to investigations, without any reasonable suspect justifying or explaining such an invasive intrusion into the private sphere. While the policies and rules regulating *familial searching* in certain States, as seen in Paragraph 2, have been adopted to limit the impact of generic and suspicionless genetic searching, genealogic databases' searches seem to be, at the moment, exempted from these strict conditions. The use by public authorities of what has been called a "fishing expedition-approach",[55] can lead police to "sneak sampling persons in the family tree [determined through the use of the genealogy database information] even though they are not suspects, simply because such samples might help expedite the investigation by eliminating potential suspect branches".[56] What emerges is the potential capability of this instrument to bypass and step over the safeguards ruling traditional DNA searches, motivated by the need to minimize the amount of sensitive data retained by law enforcement authorities in publicly run databases. Considering the great involvement of mainly innocent individuals, together with the lack of rules and regulatory oversight that characterizes this new technique, the main risk is that *forensic genetic genealogy* could be used to circumvent fundamental principles such as the presumption of innocence, freedom from unreasonable search and seizures and ban of unlimited and bulk surveillance and control.[57]

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[53] Considering the *Golden State Killer* case, it is worth mentioning that DeAngelo"s brother was convicted in California: nonetheless, his DNA profile was not collected and retained in a police-held criminal database, as his crime and conviction occurred before the Proposition 69 of California (which imposes mandatory collection and retention of DNA profile from all felons) entered into force. It is clear that if the genetic profile of DeAngelo"s brother had been uploaded in the criminal DNA database, the *familial searching* technique alone would have been successful in detecting the existent relationship between the unknown offender and his brother. But, as underlined before, the *familial searching* in criminal databases can properly work only if a genetic profile belonging to a close relative of the unidentified offender is retained. On this point, see more broadly, D. KENNET, *Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes*, cit., 114.

[54] H. L. KODY, *Standing to challenge familial searching of commercial DNA databases*, in *William & Mary Law Review*, 1, 2019, 317.

[55] D. SYNDERCOMBE COURT, *Forensic genealogy: some serious concerns*, cit., 203.

[56] E. MURPHY, *Law and policy oversight of familial searches in recreational genealogy databases*, cit., e7.

[57] "While familial searching in forensic DNA databases is framed by a series of inclusion and exclusion criteria that impose some safeguards in terms of genetic privacy, private companies have extensive databases, with few restrictions and inexistent governance. Long range familial searches in recreational DNA databases thus

3.3. The Privacy Policies established by commercial genealogy companies and the limits of the "informed consent"

In the specific US context, where this technique has firstly been used and implemented, some scholars pointed out the worst but still possible scenario: "it does not take special insight to see that law enforcement is likely to turn to genealogical databases not just to find matches in cold cases that fail to return hits in the forensic databases, but also in situations where federal or state laws expressly forbid such searches for quality control or privacy reasons".[58]

These serious concerns are strictly linked to some specific features of the *forensic genetic genealogy*. First of all, the absence – at least at the moment – of peculiar and dedicated policies and safeguards disciplining the use of this instrument is accompanied by a lack of transparency on its implementation by law enforcement authorities: in the *Golden State Killer* case, for example, only few details were initially shared by investigators about the use of genealogical databases and on the conditions and procedures followed to obtain access to data retained in these repositories. This consequently leads to a second legal as well as ethical issue: can the police upload of the unknown offender's DNA profile be considered in compliance with the privacy policy of the commercial genealogy database? And what about the "informed consent" given by the genealogy services' users? Was the possible access and search for law enforcement purposes clearly accepted by consumers? The answer to these questions entails complex considerations: the genealogy company 23andMe, in a specific "Guide for Law Enforcement" published on the company website in 2018, clearly defined a violation of its terms of service "for law enforcement officials to submit samples on behalf of a prisoner or someone in state custody who has been charged with a crime".[59] This company, similarly to MyHeritage or AncestryDNA, firmly opposes to law enforcement exploitation of its databases, unless a court order or search warrant is provided.[60]

On the contrary, GEDmatch updated its "Terms of Service and Privacy Policy" in 2018, soon after the *Golden State Killer* case became publicly debated: the company openly and clearly allows law enforcement authorities to access the database, by specifying to consumers that "while the results presented on this Site are intended solely for genealogical research, we are unable to guarantee that users will not find other uses, including both current and new genealogical and non-genealogical uses. For example, some of these possible uses of Raw Data, personal information, and/or Genealogy Data by any registered user of GEDmatch include [...] familial searching by third parties such as law

offer a way of circumventing long-established protocols in forensic DNA databases", H. MACHADO, R. GRANJA *Forensic genetics in the governance of crime*, cit., 94.

[58] E. MURPHY, *Law and policy oversight of familial searches in recreational genealogy databases*, cit., e7.

[59] See 23andMe website at https://www.23andme.com/law-enforcement-guide/.

[60] "23andMe chooses to use all practical legal and administrative resources to resist requests from law enforcement, and we do not share customer data with any public databases, or with entities that may increase the risk of law enforcement access. In certain circumstances, however, 23andMe may be required by law to comply with a valid court order, subpoena, or search warrant for genetic or personal information", in https://www.23andme.com/law-enforcement-guide/; 23andMe and AncestryDNA also publish reports communicating the amount of access-requests received and granted to law enforcement authorities for investigative purposes.

enforcement agencies to identify the perpetrator of a crime, or to identify remains".[61] Similarly and interestingly, also FamilyTreeDNA (FTDNA) declared in 2019 that "they were collaborating with the FBI and allowing them to upload DNA profiles and create accounts with the same level of access as ordinary users. Existing customers could choose to opt out of matching but this would mean that they would not benefit from the services they had paid for. It was later revealed that the FBI had already been accessing the FTDNA database for an undetermined time without the company's knowledge".[62]

Notwithstanding the different reactions of private commercial databases, reflecting the divergent positions also characterizing the civil society opinion,[63] what clearly appears as a profound and still unsolved issue is the lack of "guarantee that data shared by users actually belong to them".[64] In other words, it is difficult to imagine a way in which commercial genealogy companies could concretely enforce their positions and policies on investigators' access to their databases and assess that no violations are put in place – especially if law enforcement authorities, as happened in the *Golden State Killer* case, don't declare their identity and intentions.

Considering these weaknesses and although companies' Privacy Policies and Terms and Conditions have been, in some cases,[65] changed in order to better inform consumers, also the users' consent

[61] See the GEDMatch website at: https://www.gedmatch.com/tos.htm.

[62] D. KENNET, *Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes*, cit., 112.

[63] On this point, Kennet recalls that "a survey of 1587 US residents over the age of 18 found that the majority of respondents supported the police use of genealogy databases to identify perpetrators of violent crimes, perpetrators of crimes against children, and missing persons. The majority of respondents were not in favourof such usage to identify perpetrators of non-violent crimes. Since then, genetic genealogy databases have been used to identify the mothers of two abandoned babies and some people consider this is a step too far", D. KENNET, *Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes*, cit., 112. Also Greytak underlined that "the public is strongly in favor of the use of genetic genealogy to investigate violent crimes: GEDmatch saw a significant increase in the number of participants after the Golden State Killer arrest, and a recent survey showed overwhelming public support", E. M. GREYTAK, C. MOORE, S. L. ARMENTROUT, *Genetic genealogy for cold case and active investigations*, cit., 107. On the contrary, some authors expressed doubts on the reliability of surveys related to the public acceptance of *forensic genetic genealogy*: "as more people become familiar with the vulnerabilities of personal genetic services, opinions may shift regarding the acceptability of police access to data that are generated by and shared with these services. [...] While perceived invasions of privacy appear to be tolerable when the purpose is to catch violent or particularly depraved offenders, it seems that many would draw a line at searching their data to solve more ordinary crimes", C.J. GUERRINI, J.O. ROBINSON, D. PETERSEN, A.L. MCGUIRE, *Should police have access to genetic genealogy databases? Capturing the Golden State Killer and other criminals using a controversial new forensic technique*, in *Plos Biology*, 20, 2018, 8. See also H. MACHADO, R. GRANJA *Forensic genetics in the governance of crime*, cit.

[64] C. GUERRINI, J. ROBINSON, D. PETERSEN, A. MCGUIRE, *Should police have access to genetic genealogy databases? Capturing the Golden State Killer and other criminals using a controversial new forensic technique*, in *Plos Biology*, 10, 2018, 7.

[65] It is worth underlining that "an international review of 22 companies" and databases" policies showed that only four companies have provided additional information on how law enforcement agencies should request permission to use their services for law enforcement purposes. Two of these companies were GEDMatch and FamilyTreeDNA, two companies that permit investigative genetic genealogy – and these companies have each taken a different approach to consent. Both databases do not allow specific case-by-case consent, but rather ask for broad consent, though – more in line with dynamic consent – with the option of flexibly changing the consent settings at any time", G. SAMUEL, D. KENNET, *Problematising consent: searching genetic genealogy*

implies challenging aspects: "consumers of genealogy tests now have to confront the tension between the need to protect their own privacy and that of their close and distant relatives, and their strong desire to use this information for their own genealogical research. It is for them to choose between the two, but they need to do so on an informed basis"[66] In order to give a truly informed consent, users should be clearly made aware that their decisions "to contribute their own genetic information inadvertently exposes many other across their family tree who may not be aware of or interested in their generic relationships going public".[67] Differently from the *familial searching* of national DNA databases, which implies that no free consent has been given by convicted criminals or arrestees – whose DNA profile is mandatorily included in the police-held database –, the choice to upload a genetic profile to a genealogy services' website is voluntary but, in this specific case, not strictly "personal" because of the effects and "indirect" involvement produced over – even distant and mainly unaware – relatives.[68]

Following these considerations, some scholars started talking about "generational consent", as a new form of consent including "more than just the individual in decisions about participating in genetic investigations".[69]

This dynamic and still open debate on privacy and data protection concerns linked to the development of genetic genealogy investigations, highlights how this new technique imposes a serious reconsideration and re-thinking of the "traditional" idea of consent. If this instrument is still recognized of key importance, especially in contexts, such as the US one, where privacy and data protection legislations are not fully in place,[70] the peculiarities and specific challenges posed by

databases for law enforcement purposes, in *New Genetics and Society*, 2020, https://bit.ly/3dFvebx, 5, recalling a survey of S. SKEVA, M. LARMUSEAU, M. SHABANI, *Review of policies of companies and databases regarding access to customers" genealogy data for law enforcement purposes*, in *Personalized Medice*, 2, 2020, 141-153.

[66] D. SYNDERCOMBE COURT, *Forensic genealogy: some serious concerns*, cit., 204.

[67] S.M. FULLERTON, R. ROHLFS, *Should police detectives have unrestricted access to public genetic databases?*, in *Leapsmag*, 23 July 2018.

[68] Dangers are even more profound if we consider how difficult it is for consumers to fully understand and properly evaluate the risks for their privacy and data when reading usually complex services" privacy policies (especially if online): a truly informed and free consent should be promoted though transparent, clear and easy terms and conditions, giving the consumer a concrete idea of the perils and possible side effects deriving from his/her approval. Moreover, it should be properly considered that "consumers have a tendency towards inertia, particularly when decisions are complex, meaning that they are unlikely to change their opt-in preferences related to consent on the website", G. SAMUEL, D. KENNET, *Problematising consent: searching genetic genealogy databases for law enforcement purposes*, in *New Genetics and Society*, 2020, https://doi.org/10.1080/14636778.2020.1843149, 5. This point underlines the problems and weaknesses related to the use of "opt-out" policies.

[69] "Traditional informed consent reflects individualistic decision-making. We argue that it is time to think of consent in broader terms, as a discussion that, when involving genetic information, goes beyond the individual and asks all parties to think about and involve the broader family and biological relatives", S.E. WALLACE, E. GOURNA, V. NIKOLOVA, N. SHEEHAN, *Family tree and ancestry inference: is there a need for a "generational consent"?*, in *BMC Medical Ethics*, 16, 2015, https://doi.org/10.1186/s12910-015-0080-2.

[70] In US, the use of genetic data and genetic privacy are regulated by two federal laws: the Health Insurance Portability and Accountability Act of 1966 (HIPAA) and the Genetic Information Non-discrimination Act of 2008 (GINA). Some States have approved general data protection laws, providing safeguards also to sensitive data, such as genetic and biometric data or have adopted specific provisions disciplining "genetic privacy". A federal general legislative framework regulating data protection, similarly to the EU Regulation 2016/679, is not in

genealogy forensic must be carefully taken into account: the "traditional" individualistic dimension of consent less suits the privacy issues related to genealogy databases.[71]

All the challenges identified in this paragraph provide a clear picture of the complex concerns and challenges highlighted by lawyers, academics, civil society, geneticists and law enforcement authorities: the still open and unanswered doubts and questions – concerning potentialities, efficiency,[72] side effects and risks, privacy, safeguards and limits to be put in place – call for a profound discussion able to result in a clear and crucial intervention of legislators and policymakers.

4. How to avoid "genetic surveillance": paving the path towards a profound guarantee of "genetic privacy"

4.1. The risks of a "universal database": some timid attempts of regulatory answers

The critiques and the serious ethical and legal concerns emerged in the aftermath of the *Golden State Killer* case have not prevented US law enforcement authorities to solve many other cold or active cases thanks to *forensic genetic genealogy.*[73] This tendency seems to confirm that, "far from being a forensic anomaly, the public genetic search is quickly on its way to becoming routine procedure".[74]

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place. On the gaps of the existent legislative discipline and the problematic approach adopted by some federal and State Courts, see R.M. HENDRICKS-STURRUP, A. PRINCE, *Direct-to-consumer genetic testing and potential loopholes in protecting consumer privacy and non discrimination*, in *JAMA*, 19, 2019, 1869 ff.; S. LUND, *Ethical implications of forensic genealogy in criminal cases*, in *The Journal of Business, Entrepreneurship & the Law*, 2, 2020, 203 ff.

[71] See also N. SCUDDER, *Privacy and the search for suspects using forensic genetic genealogy*, in *Privacy Law Bulletin*, 5, 2020, 78-81; more generally on consent, A.M. FROOMKIN, *Big Data: destroyer of informed consent*, in *Yale Journal of Health Policy, Law and Ethics*, 3, 2019, 30-54.

[72] "While the commercial autosomal DNA relative-matching tests have essentially been validated by usage by millions of genealogists, the methodologies have not been validated for forensic use. Forensic samples are likely to be degraded, producing a large a number of no calls, and it is not known what impact this will have on the relationship predictions. The proprietary techniques used by Parabon, the DNA Doe Project and the other companies are still experimental and have not been subjected to peer review, creating concerns about transparency and accountability", D. KENNET, *Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes*, cit., 109.

[73] On this point, see H. MACHADO, R. GRANJA *Forensic genetics in the governance of crime*, cit., who also recall Y. ERLICH, T. SHOR, I. PE"ER, S. CARMI, *Identity inference of genomics data using long-range familial searches*, in *Science*, 6415, 690-694; but also E.M. GREYTAK, C. MOORE, S. L. ARMENTROUT, *Genetic genealogy for cold case and active investigations*, cit.

[74] C.J. GUERRINI, J.O. ROBINSON, D. PETERSEN, A.L. MCGUIRE, *Should police have access to genetic genealogy databases? Capturing the Golden State Killer and other criminals using a controversial new forensic technique*, cit., 9. Doubts on a possible extensive employment of this techniques were initially expressed by some scholars, mainly because of the elevate costs required in terms of time, money and human resources (see E. MURPHY, *Law and policy oversight of familial searches in recreational genealogy databases*, cit., e7.) Nonetheless, technological and scientific advancements (such as in the field of algorithms and AI instruments) could make the implementation of this instrument more and more easier in the future.

Similar to what happened with regards to the *familial searching* technique and although DNA searches of third-party databases remain mainly an "uncharted" territory, in recent years we are witnessing some first timid attempts to discipline the use of forensic genealogy.

In January 2019 the US Department of Justice approved an interim policy,[75] establishing useful guidelines and case-criteria: "investigative agencies may initiate the process of considering the use of forensic genetic genealogy searches (FGGS) when a case involves an unsolved violent crime (homicide, sex crime) and the candidate forensic sample is from a putative perpetrator, or when a case involves what is reasonably believed by investigators to be the unidentified remains of a suspected homicide victim. In addition, the prosecutor may authorize the investigative use of FGGS other than violent crimes [...] when the circumstances surrounding the criminal act(s) present a substantial and ongoing threat to public safety or national security. Before an investigative agency may attempt to use FGGS, the forensic profile derived from the candidate forensic sample must have been uploaded to CODIS and subsequent CODIS searches must have failed to produce a probative and confirmed DNA match" (point V); the prosecutor should assess that the genealogy forensic is a necessary and appropriate step to develop investigative leads at that stage of the investigation (point VII). Specific rules on the relationship between investigators and genealogy services and databases are established, providing that "investigative agencies shall identify themselves as law enforcement to genetic genealogy services and enter and search profiles only in those services that provide explicit notice to their service users and the public that law enforcement may use their service sites to investigate crime or identify unidentified human remains" (point VII).

By determining limits and rules, these guidelines certainly go in the direction of stronger restrictions and comprehensive safeguards; nonetheless the interim policy still "have room for improvement and still leave the door open for troubling privacy violations":[76] the provisions apply only to the Department of Justice agencies – so that State and local law enforcement authorities are excluded from the scope of application of this document – and the numerous exceptions risk to legitimize discretionary decisions. Terms such as "threat to public safety or national security", allowing for *forensics genealogy* out of the specific serious crimes' cases listed in the policy, could be extensively interpreted and applied. The "explicit notice" law enforcement agencies are required to give to private genealogy databases is an important safeguard, prohibiting what already happened in the past (the upload of a DNA profile by investigators without disclosure of their status and their purposes). But this doesn't guarantee a complete users' protection: the unclear and often not-understandable privacy conditions provided by the genealogy services don't allow for a fully informed consent of the consumer; on the contrary, requiring "an opt-in approach, whereby law enforcement only receives access when a user actively gives permission, would ensure that users approve the site's policy"[77] or the changes applied over the time.[78] Furthermore, no obligation to

[75] Available at https://www.justice.gov/olp/page/1204386/download.

[76] J. SCHWAB, *New DOJ policy gives genealogy website users weak privacy protections from law enforcement*, in *Harvard Civil Rights – Civil Liberties Law Review*, 3 October 2019, https://harvardcrcl.org/new-doj-policy-gives-genealogy-website-users-weak-privacy-protections-from-law-enforcement/.

[77] J. SCHWAB, *New DOJ policy gives genealogy website users weak privacy protections from law enforcement*, cit.

ISSN 2284-4503

notify affected users is specified in the approved guidelines nor a warrant, based on probable cause, is requested; the policy doesn't even answer the privacy concerns linked to the "individual" nature of the consent and to the consequent need to protect not only the consenting user but also his/her relatives. Consequently, if this interim policy is a first meritorious attempt to address the challenges posed by this new investigative tool, it should nonetheless leave space for a profound and comprehensive revision, able to re-consider all the problematic legal and ethical aspects emerged from the public debate.[79]

The evolution of the *forensic genetic genealogy* in the US is carefully followed also in Europe: in September 2020 the UK Government published a Report of the Biometrics and Forensics Ethic Group,[80] focusing on the feasibility of such technique in the UK context. In the conclusions provided by the advisory group a very cautious approach emerges: "the legality and necessity of police use of genetic genealogy in the UK would need to be clearly established with reference to Art. 8 ECHR and the Human Rights Act 1988. The approach should be used if it can be shown to be based on clear evidence, verified by an independent body, that the established methods already in use for these law enforcement purposes are no longer adequate or effective. Otherwise, the use of any such novel processes would not meet the tests of necessity and proportionality. This would make the legality of using such novel processes highly suspect. [...] Legislation for the transmission, length of retention, and destruction of the sample, profile and collected genealogical data would be needed".[81] In requiring prior and comprehensive rules and safeguards, the Group seems to question the legitimacy and the concrete utility of this technique in UK, also underlining that "UK already has one of the most efficient DNA databases in the world and conventional methods, with appropriately applied familial searches, will identify the bulk of perpetrators".[82]

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[78] In this sense, it is worth noting that in May 2019 "GEDmatch revised its policy to an active "opt in", where consumers had to actively agree to be included in any searches done by government agencies", S. LUND, *Ethical implications of forensic genealogy in criminal cases*, cit., 202.

[79] S. LUND, *Ethical implications of forensic genealogy in criminal cases*, cit., 207, recalling P. ST. JOHN, *DNA genealogical databases are a gold mine for police, but with few rules and little transparency*, in *Los Angeles Times*, 24 November 2019, https://www.latimes.com/california/story/2019-11-24/law-enforcement-dnacrime-cases-privacy .

[80] "The Biometrics and Forensics Ethic Group is an advisory group non-departmental public body, sponsored by the UK Home Office. The group provides advice on ethical issues in the use of biometric and forensic identification techniques such as DNA, fingerprints, and facial recognition technology", https://www.gov.uk/government/publications/use-of-genetic-genealogy-techniques-to-assist-with-solving-crimes/should-we-be-making-use-of-genetic-genealogy-to-assist-in-solving-crime-a-report-on-the-feasibility-of-such-methods-in-the-uk-accessible-version.

[81] See the Report at https://www.gov.uk/government/publications/use-of-genetic-genealogy-techniques-to-assist-with-solving-crimes/should-we-be-making-use-of-genetic-genealogy-to-assist-in-solving-crime-a-report-on-the-feasibility-of-such-methods-in-the-uk-accessible-version.

[82] Notwithstanding the open debate, some authors have already highlighted that "a small convenience sample pilot study has already demonstrated the method would work in the UK setting", G. SAMUEL, D. KENNET, *Problematising consent: searching genetic genealogy databases for law enforcement purposes*, in *New Genetics and Society*, 2020, https://doi.org/10.1080/14636778.2020.1843149, 3; see also J. THOMSON, *An empirical investigation into the effectiveness of genetic genealogy to identify individuals in the UK*, in *Forensic Science International: Genetics*.

Regardless of the different possible views and approaches, what seems to be uncontroversial is, on the one side, an increasing tension towards an extensive use of DNA analysis in the law enforcement field and, on the other side, a growing awareness of the serious challenges deriving from these techniques.[83]

4.2. How to resist the temptation of "seeing into the life of citizens": prompting a thoughtful and pondered debate

Even if not always totally or *per se* decisive,[84] the development of new forensic DNA techniques has enabled "a new wave of crime-solving technology",[85] with particularly positive effects on the capability to solve cold cases, when all the other possibilities revealed a dead end. As Syndercombe Court underlined, the *familial searching* of DNA criminal databases has been considered, since the beginning, "a 'quantum leap' in forensic identification, and is made even more significant today by the use of genealogical databases".[86] Both these investigative tools move "the locus from individualization, that is, identification of specific individuals, towards collectivization [...], by clustering 'suspect' populations which share biological links and genetic ancestry".[87] By doing so, these innovative instruments have been extensively criticized for their capability to expose innocent people to life-long surveillance[88], to exacerbate already existent racial inequalities and discriminations and to consequently debunk 'genetic privacy' safeguards. What is feared the most is the possible detrimental shift to "a *de facto* universal database", especially through the use of genealogy connections:[89] this will conduct to a significant expansion of "the scope and impact of genetic surveillance",[90] by "constructing suspicion as collective".[91] The use of private companies'

[83] In 2008, the ECtHR, in the already recalled *S and Marper v. UK* decision, recognized the dangerous tendency to allow "modern science techniques in the criminal-justice system [...] at any cost and without carefully balancing the potential benefits of the extensive use of such techniques against important private life interests", para. 112.

[84] Using these research methods does not always assure a result: "while there certainly will be more announcements of cases solved using this new technique, there are many more cases where identification has not yet been possible, due to the wide variety of complications present in these investigations", E. M. GREYTAK, C. MOORE, S. L. ARMENTROUT, *Genetic genealogy for cold case and active investigations*, cit., 103.

[85] R. WICKENHEISER, *Forensic genealogical searching and the Golden State serial killer*, in *Forensic Science International*, 1, 2019, S9.

[86] D. SYNDERCOMBE COURT, *Forensic genealogy: some serious concerns*, cit., 204, recalling E. MUPRHY, *Relative doubt: familial searches of a DNA database*, in *Michigan Law Review*, 109, 2010, 291-348.

[87] H. MACHADO, R. GRANJA, *Forensic genetics in the governance of crime*, cit., 86.

[88] S. KRISMKY, T. SIMONCELLI, *Genetic justice: DNA data banks, criminal investigations and civil liberties,* New York, 2011.

[89] E. MURPHY, *Law and policy oversight of familial searches in recreational genealogy databases*, cit., e7. According to the author, "essentially everyone will be a police database now". Expressing a pessimistic view, Murphy forecasts that "the prevalence of genealogical DNA databases searches will begin to infect the debate about the use of governmental databases, and prompt the loosening of existing regulations rather than the enhancement of the regulatory architecture for genealogical searches", e7.

[90] A similar expression was significantly used by Justice Scalia in his Dissenting Opinion in the abovementioned *Maryland v. King* decision (*supra* note 11): in that case – concerning the possibility to collect DNA samples from arrestees – Justice Scalia warned against the perils of a "genetic panopticon", para. 1900.

[91] C. MACHADO, R. GRANJA, *Forensic genetics in the governance of crime*, cit., 92 and 99.

databases, less regulated and controlled compared to national and publicly-led databases, brings an additional layer of complexity.[92]

The seriousness of the underlined risks and the rapidly increasing use of sophisticated but highly intrusive investigative techniques, mainly in the absence of specific regulatory frameworks, prompts for a thoughtful and pondered legislative discussion, before these instruments become widely applied. Accordingly, a comprehensive set of rules should be determined, on the basis of a careful risk-assessment: with regards to *forensic genetic genealogy*, "there must be a process to ensure genealogical searching is conducted properly scientifically and from a public policy perspective. There should be transparency of policies, procedures and documentation to guide and demonstrate appropriate use".[93] A dedicated training of law enforcement authorities, illustrating a proper and correct application of the new techniques, should be followed by an exhaustive discipline of privately-run genealogy services, also strengthening the effectiveness of a truly informed consent on the consumers' side: it can be, for example, imposed to genealogy companies and databases to clearly and unambiguously inform users about the risk to expose their genetic privacy and that of their relatives to the access of law enforcement agencies for investigative purposes, extensively explained and priorly determined. Company reports on the factual access by investigators should be a recommended practice. Moreover, specific policies and laws should be approved in order to establish precise limits to the employment of both *familial searching* and *forensic genetic genealogy*: determining what kind of crimes could allow for familial or genealogy searching in private databases, or what kind of requests should law enforcement authorities present, in a transparent way, to the genealogy companies, are of paramount importance to set well-defined safeguards able to minimize abuses and avoid an extensive recourse to invasive techniques, which should be considered the last possible resort. The conditions that justify the implementation of these investigative tools, virtually capable of targeting a vast number of innocent people, should be determined according to the principle of proportionality, necessity and data minimization. These considerations should inspire and guide legislators and policymakers to rapidly move towards efficient and comprehensive regulatory answers.

[92] Divergent points of view are expressed on this difficult topic: some authors consider that if a person, based on correct information, voluntarily decides to upload his/her DNA profile on a genealogy website, there are no reasons to prevent police from employing these data (E. M. GREYTAK, C. MOORE, S.L. ARMENTROUT, *Genetic genealogy for cold case and active investigations*, cit.). On the contrary, Kody affirms that "allowing a company to analyse one"s DNA for medical or ancestry purposes does not do away with the protection all Americans have to be free from unreasonable and unwarranted government intrusion", L. KODY, *Standing to challenge familial searching of commercial DNA databases*, cit., 318.

[93] R. WICKENHEISER, *Forensic genealogy, bioethics and the Golden State Killer case*, cit., 123. Similarly, Berkman, Miller and Grady affirmed that "a commitment to transparency is extremely important. Authorities apparently are reluctant to admit that they use forensic DNA searching, despite the fact that most states do so. If law enforcement is using this technology, the adoption of formalized standards and mechanisms of accountability is appropriate. The limits of DNA evidence also suggest that restrictions should be placed on its use. We recommend using forensic genealogy as an investigative tool rather than a primary source of evidence of criminal wrongdoing. Likewise, justice concerns might warrant limiting criminal genealogy searching to cold cases involving crimes in which other investigative methods have failed", B. BERKMAN, W. MILLER, C. GRADY, *Is it ethical to use genealogy data to solve crimes*, in *Annals of Internal Medicine*, 5, 2018, 334.

In this context, the question that should be posed at the very basis of every consideration and decision should be "to what extent can the rights of the innocent general public and relatives of the committer of a crime be infringed upon by examining their genetic data to identify the crime perpetrator and thereby prevent future crimes and improve public safety?".[94] Although addressing this question and determining a correct balance-point will probably be imperfect and non-final,[95] the necessary answers cannot be left to the consent of users or to the privacy policies set by private companies, and should, on the contrary, be properly managed by public policies and laws.

In conclusion, the rising implementation of *familial searching* and *genetic genealogy* forensics exemplify the diffuse public authorities' desire to fully exploit the potentialities of very sensitive and personal data together with the opportunities represented by new technological tools or procedures. The tendency to collect, retain, access and employ a great amount of data for the sake of security or efficiency of public services is visible in many other fields, from the mandatory retention of telecommunications' metadata for public and national security purposes to facial recognition technologies as crime-fighting tool, from biometric identification systems necessary to access fundamental welfare services, to automated risk management tools used to detect tax and welfare frauds:[96] these trends draw a dangerous shift towards over-surveillance and the creation of societies in which citizens are subjects to control and intrusion in their more intimate sphere, also through the use of very unique and sensitive data.

As former ECtHR Judge Pettiti clearly stated, back in the 1980s, "the danger threatening democratic societies [...] stems from the temptation facing public authorities to see into the life of citizens".[97] This widespread temptation – fuelled by the increasing "datification", digitalization and technological progress and possibly able to undermine, at their very basis, fundamental rights' guarantees and safeguards – must be seriously and rapidly tackled, in all its expressions, by civil society, scholars, legislators and Courts. This paper's ambition – and hope – is to help keep such a vital debate alive.

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[94] R. WICKENHEISER, *Forensic genealogical searching and the Golden State serial killer*, cit., S9.

[95] As brilliantly stated by Suter, with regards to *familial searching*, "in some ways the conflict seems insoluble. Proponents and opponents of familial searching [but the same is true for genetic genealogy forensic] are both fighting the "good fight". Both are motivated by defensive postures. Proponents want to fight crime; opponents want to fight violations of civil liberties. When each side is so deeply passionate about its underlying goals, it becomes difficult not only to find a compromise, but even to agree upon a common approach to resolving this and other difficult dilemmas. In short we face the challenge of there being a plurality of important values, some of which collide. How do we handle this collision and the possibility that some of the values may have to give away in certain contexts?", S. SUTER, *All in the family: privacy and DNA familial searching*, cit., 375.

[96] See for example the legal challenges emerged from the "data retention regime" in the EU as well as the long and complex ECJ "data retention saga"; see also the controversial use of Syri: this program, adopted in the Netherlands and aimed at detecting tax frauds through the automated analysis of data collected and retained by public agencies, was declared unlawful by the District Court of the Hague because of its lack of transparency and disproportionate interference with citizens" private life. The debate is still open in Ireland and France for the adoption of automated biometric identification systems aiming at granting citizens" access to public welfare services; similarly, the use of facial recognition for law enforcement purposes, based on the collection and comparison of sensitive data belonging to mainly innocent citizens, has been challenged before UK Courts.

[97] ECtHR (2 August 1984), *Malone v. UK*, n. 8691/79, Judge Pettiti Concurring Opinion, para. 38.

ISSN 2284-4503

Special issue

Forensic genetics: The evolving challenge of DNA cross-border exchange

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ABSTRACT: Forensic genetics has significantly reshaped the investigations' methods and simultaneously brought numerous ethical and legal challenges, especially in the area of DNA data exchanges. By retracing the history of DNA cross-border exchanges within the so-called Prüm framework, this paper aims to underline not only the current inhomogeneity of Member States' legislations in this complex field but also the existing inefficiencies in the implementation and application of the "Prüm system". On this basis, potential developments and possible reform solutions are analysed, with the purpose of explaining and reflecting on the opportunities and risks enshrined in the so-called Next Generation Prüm.

KEYWORDS: DNA; DNA databases; genetic forensics; Next Generation Prüm

SUMMARY: 1. The DNA on trials: what has been uncovered and what remains hidden – 2. Learning from experience. The history of DNA cross-border exchanges and their expansion – 3. The inhomogeneity of Member States' legislations and the difficult implementation of the "Prüm system" – 4. The uncharted territory of Next Generation Prüm.

1. The DNA on trials: what has been uncovered and what remains hidden

The outbreak of DNA analysis as forensic evidence in criminal investigations and Courts has brought important legal and ethical issues.[1] Indeed, the deployment of such new methodology has made crystal clear that the "circulation" of genetic data is linked not only has undeniable advantages but also potential risks related to the use of genetic information. Moreover, methods of collection, retention and deletion of data, both within the European Union and around the world, may differ significantly according to different domestic legislations; therefore, many difficulties arise with regards to the use and exchange of such data.[2] When we discuss forensic evidence related to genetic data, it is important to remember that the perspective may shift beyond the specific individuality of a person and become ultra-personal. Thus, it is essential to understand how a new axiological arena has taken shape and how different rights and freedoms, from personal

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[1] On this point, see a recent and comprehensive contribution: H. MACHADO, R. GRANJA, *Forensic Genetics in the Governance of Crime*, Singapore-Braga, 2020.

[2] For a broader reflection on this specific issue, see: L. SCAFFARDI, *Giustizia genetica e tutela della persona. Uno studio comparato sull'uso (e abuso) delle Banche dati del DNA a fini giudiziari*, Milano, 2017.

ISSN 2284-4503

freedom to privacy, from the right to health to informed consent, could be balanced with other subjective legal positions.

This paper aims to provide an evolving discussion on how the use of DNA fingerprint has reshaped the debate on some of the rights involved. Courts and judges have played, in this context, an important role, given that judgments have become essential to building a new interpretation of the subject, inherently linked with its practical dimension and with relentless technological progress.

The impulse brought by rulings has been essential to provide interpretative parameters guaranteeing individual and ultra-personal rights. New and interesting developments are nowadays characterising this subject, which is still far from being untangled. Possible enhancement in the use and exchange of genetic data and non-genetic data may in fact be found in the innovations proposed in the Next Generation Prüm, which will be analysed in the following paragraphs. It is therefore evident, in such a scenario, that genetic data, as forensic evidence, represents an open and always evolving challenge.

2. Learning from experience. The history of DNA cross-border exchanges and their expansion

The use of DNA for identification purposes dates back to 1985 when Alec Jeffreys paved the way for this new methodology,[3] analysing and comparing the genetic material found on a crime scene with the one of a suspect. Thanks to the continuous enhancement of molecular technology techniques, it became possible to implement DNA repositories, which allow an electronic comparison of the data contained within them. Obviously the creation of specific databases has further increased the possibilities to solve police investigation, turning genetic tests into a real "operational system": in 2007 Grimm underlined that the USA genetic database[4] was able to analyse 100.000 profiles in 500 microseconds.[5] In the European context, it can be pointed out that the total number of identifiable

[3] See A.J. JEFFREYS, V. WILSON, S.L. THEIN, *Individual-Specific "Fingerprints" of Human DNA*, in *Nature*, Vol. 316, 1985.

[4] For a reconstruction of the debate, occurred in the USA, around different methods that led to an increasing expansion of the central database, see J. D. ARONSON, *On trial! Governing forensic DNA technologies in the USA*, in R. HINDMARSH, B. PRAINSACK (a cura di), *Genetic Suspects: Global Governance of Forensic DNA Profiling and Databasing*, Cambridge, 2010, 254 ff.

[5] D.J. GRIMM, *The demographics of genetic surveillance: familial DNA testing and the hispanic community,* in *Columbia Law Review*, Vol. 107, 2007, 1169. In order to understand the swirling increase (updated to December 2020) of the DNA profiles' number currently at the disposal of USA authorities at both national and federal level see: https://www.fbi.gov/services/laboratory/biometric-analysis/codis/ndis-statistics. Within the vast bibliography on this topic, particular attention should be given to the following contributions and the references cited therein: S. KRIMSKY, T. SIMONCELLI, *Genetic Justice. DNA Data banks, Criminal Investigations and Civil Liberties*, New York, 2011; M. TAYLOR, *Genetic Data and the Law. A critical Perspective on Privacy Protection*, Cambridge, 2012; K. J. STROM, M. J. HICKMAN, *Forensic Science and the Administration of Justice,* in *Critical Issues and Directions*, London, 2015.

persons' profiles stored in national databases amounts to approximately 11,5 millions, along with 1,6 million of data that refer to non-identifiable persons.[6]

The potential of such data should be considered not only with regard to their possible use at national level but also as important information to be exchanged between Member States in order to improve the investigative cooperation.

In the last few decades, the rise and development of forensic genetic databases have been strongly affected by the possibility of cross-border exchange of data[7] (within and beyond EU borders) and by the constant technological process, which has led to increasingly pervasive methods of production, collection and retention of data.[8] On the other hand, crimes have progressively taken on a cross-border dimension –international terrorism, drug trafficking or human smuggling and, moreover, irregular migration and organised crime. It has become more and more essential to provide law enforcement authorities with investigative tools that can cross borders and enhance information exchange and transnational cooperation.

In this context, for what specifically in relation to genetic data, DNA exchange systems have proliferated; such systems can be defined as mechanisms of DNA cross-borders exchange (which allow for the comparison and matching of DNA profiles) that can assume different and constantly evolving dimensions and characteristics.[9]

An interesting and recent attempt at systematization[10] has shown that there are different types of *exchange systems,* among which particular attention may be given to*:* (1) *international database,*

[6] European data provided by European Network for Forensic Science Institutes (ENFSI) in 2017 and available at www.enfsi.eu. For a comprehensive study about data collection in various continents see INTERPOL, see *Global DNA Profiling Survey Results*, 2019.

[7] In order to understand such developments, it is interesting to see the scientific study developed between 2011 and 2015, reporting data concerning DNA profiles' exchange among EU Member states. This study represents the first systematic analysis in this field, based on official statistics drafted pursuant to the adoption of the so-called Prüm system. It clearly shows that the difficulty of finding statistic data, which is still a complicated and outstanding problem, is a main issue. Another challenging topic is related to the fact that, still nowadays, it is very complex to properly establish the concrete results deriving from implemented policies and to consequently guarantee transparency and accountability vis-à-vis citizens. Nonetheless, the study underlines the implementation of one-to-one exchanges between some Member states, among which Belgium, France, Netherlands and United Kingdoms. See F. SANTOS, H. MACHADO, *Patterns of exchange of forensic DNA data in the European Union through the Prüm system*, in *Science & Justice*, Vol. 4, no. 57, 2017, 307-313.

[8] It is essential to specify that the notion of "genetic data", in this context, needs to be interpreted in a specific way: in particular for what concerns the information exchange system among Member States for investigative purposes, "genetic data" refers to the information resulting from the non-coding part of the DNA. Data to be exchanged do not contain information that could lead to the direct identification of a subject, at least in the first phase of the exchange system, which will be analysed below.

[9] This constant evolution of cooperation and data exchange system is influenced by both a growing number of States that decide to implement national DNA databases and a continuous technical-scientific progress that allows for the development of new research methods and innovative investigative systems based on genetic data. Consider, as an example, the so-called familial searching technique, analysed in this Journal in G. FORMICI, *From "familial searching" to "forensic genetic genealogy": new frontiers – and challenges – of DNA analysis in criminal investigations*.

[10] A.O. AMANKWAA, *Trends in forensic DNA database: transnational exchange of DNA data*, in *Forensic Sciences Research*, no. 1, 2019.

such as the Interpol database;[11] (2) interrelated and connected national DNA databases, that can be found in the EU system based on the Prüm Treaty; (3) bilateral or multilateral agreements concerning the exchange of genetic data following the request of a national authority.[12]

Given that one of the main potentials uses of databases is the possibility to compare data and exchange information, it goes without saying that a national system should not be considered on its own but, rather, regarded within the general context of European rules, that strongly affect domestic choices.

The value of these tools is evident[13] but it is, at the same time, important to take into proper consideration the risks that may be inherent to the sharing and exchanging of sensitive data like genetics. The invasiveness of these operations is significant, especially with regards to privacy and data protection, therefore it is essential to think carefully about the need to provide rules that fully guarantee the rights of the individual, in the context of sharing and exchanging information.

Consequently, it is evident that the main challenge that national and supranational legislators are facing is finding the correct balance between security[14] and respect of privacy and data protection rights.[15] Indeed, the former calls for fully operating systems of collection, retention, access and exchange of data while the latter requires conditions able to guarantee that every interference in the personal sphere of the data subject is proportionate to the purpose being pursued.

Facing such a complex challenge, the European Union has demonstrated great attention to this, deciding to "Europeanise" the provisions of the Prüm Treaty (i.e., encompassing them into its normative framework), which namely, DNA exchange, fingerprint data and information about the registration of vehicles.

In doing so, the EU imposed its Member States to adopt national legislation governing collection and exchange of genetic data for judicial purposes, in compliance with supranational criteria. Member States were required to develop national databases, as an essential prerequisite of comparing and matching different profiles coming from other Member States' authorities and for the subsequent exchange of information of the identified suspect.

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[11] Built in 2002, this database contains more than 180.000 genetic profiles, gathered with the cooperation of more than 84 Member States: "Police can submit a DNA profile taken from offenders, crime scenes, missing persons and unidentified human remains, with a search result provided within minutes. Our database has enabled investigators around the world to link offenders to different types of crime including rape, murder and armed robbery. There is no nominal data attached to the profile, which is submitted in the form of an alphanumerical code. Member countries retain ownership of the information, in line with our rules on the processing of data. Countries can also choose which other countries they share their data with", https://www.interpol.int/How-we-work/Forensics/DNA

[12] This category includes the important bilateral agreements concluded between USA and other Countries around the world, which define specific conditions for genetic data exchange (The agreement with Italy dates back to May 28th 2009).

[13] See P. GILL, *National DNA Databases, Strength of Evidence and Error Rates*, in P. GILL (ed), *Misleading DNA Evidence*, Cambridge, 2014, in particular 81 ff.

[14] See L. SCAFFARDI, *Banche Dati del DNA e scambio internazionale fra esigenze securitarie e tutele dei cittadini,* in L. SCAFFARDI (ed), *La banca Dati italiana del DNA. Limiti e prospettive della genetica forense*, Bologna, 2019.

[15] These rights are ensured, at the EU level, within the context of the Charter of fundamental rights of the European Union, incorporated as additional protocol by articles 7 and 8 of the Lisbon Treaty.

Consequently, it is obvious that the creation of a national databases' network, formally separated but tied together by cooperation obligations, had a huge impact on the domestic system of each Member state. It should however be pointed out that the supranational legislation has left a certain degree of discretion to national legislators, which resulted in different approaches. Thus, it is essential, in a comparative perspective, to reflect on the similarities (i.e. homogeneity of legislative choices) and/or on the differences that characterise domestic strategies that, within a continuous dialogue with the EU level and among Member States, will affect the capability and the efficiency of the whole system.

3. The inhomogeneity of Member States' legislations and the difficult implementation of the "Prüm system"

The inclusion of specific provisions regulating the use and exchange of genetic data for judicial purposes, within the EU legal framework, was a long and troubled journey. In 2005 seven Member States decided to sign the so-called Prüm Treaty, open to all other Members wishing to join, with the aim of encouraging the implementation of a system of collection, access and exchange of data, such as DNA, fingerprints and information about registration of vehicles, in order to enhance the mechanisms of police cooperation. The Treaty stemmed from the need to fight criminal activities in a more efficient way that were, at that time, rapidly expanding (e.g., terrorism, illegal immigration, organised crime). By understanding the importance of that agreement and being willing to include it in the EU legal framework, the Council adopted, in 2008, the Decisions 2008/615/JHA and 2008/616/JHA, after a long and trouble *iter*, often characterised by setbacks and concerns expressed by the EU institutions involved.[16] These Decisions replicated the provisions of the Prüm Treaty[17] and provided Members states with the possibility of "sending" a DNA profile to another Member state if the identification process didn't result in a match, at national level within their own DNA database. The ither Member States, receiving the DNA profile, can subsequently verify if they can find a matching profile within their own DNA national databases. This first "hit/no hit" phase is fully-automated and is carried out by the so-called national points of contact; it does not require a proper exchange of personal data but rather a mere comparison – by means of "silent codes" – between the profile sent by the requesting Member state and the ones collected in all other national databases within the EU. Only in the case of a positive response (hit), does a second phase take place and the personal information related to that profile can be requested so that it becomes possible to trace the identity of the person to whom that profile belongs. This procedure, dived into two separate steps, allows for a greater speed in the first phase and guarantees a higher level of data protection, by imposing a disclosure of personal data only in the second phase and only if the automated operation of comparison and matching resulted in a positive outcome. The abovementioned exchange of data

[16] Both the European Data Protection Supervisor and European Parliament expressed doubts with regard to the decision and the employed normative source and proposed some amendments to the text draft elaborated by the Council.

[17] Decision 2008/615/JHA, point 10.

can only take place after the implementation of the EU Decisions at national level:[18] it is necessary to set up national databases, through which the operations of checking will be conducted, along with the establishment of "Contact points" and formalised ways of requesting and replying to other Members States' demands. All these requirements need to be implemented by national legislators who are also required to define the criteria and conditions for the entry of genetic data (samples or profiles) into the national databases.[19]

Some harmonizing measures were also introduced with the Council Decisions: for example, legislators are asked to comply with some specific common requirements,[20] such as defaulting European standards (ESS; ISSOL). It is moreover compulsory to conform to others technical standards: with regard to the processing of the data that will be exchanged, each Member State must guarantee a certain degree of data protection within its domestic framework.[21]

This drive to harmonize the system, is reaffirmed by the fact that the Council has implemented a control procedure which allows them to check if Member States are complying to European standards and thus gives them to the power to authorize or deny access to the national databases' network.

Although the Union has repeatedly underlined the necessity to reach a complete harmonisation of national provisions, there are still many disparities among Member States concerning both the correct implementation of the abovementioned Decisions and the normative choices in this field. Such a complex scenario also emerges from the Fifth progress report towards an effective and genuine Security:[22] "In the area of information exchange between Member States, the Prüm Decisions of 2008 introduced procedures for fast and efficient data exchanges among Member States by laying down rules and providing a framework to allow Member States to search each other's DNA analysis files, fingerprint identification systems and vehicle registration data bases. Prüm was a tool that helped French investigators after the Paris terrorist attacks of November 2015. Considerable progress has been made in the implementation of Prüm in recent months with increasing volumes of data exchange. However, a number of Member States have still to implement the Decisions almost a

[18] Art. 25(2), Decision 2008/616/JHA.

[19] It is up to national legislators to adopt national laws that define conditions and criteria for the data processing and exchange, conditions that requesting Member States must always comply with (art. 26(1), Decision 2008/615/JHA).

[20] "Member States shall observe common technical specifications in connection with all requests and answers related to searches and comparisons of DNA profiles, dactyloscopy data and vehicle registration data. These technical specifications are laid down in the Annex to this Decision", art. 3, Decision 2008/616/JHA.

[21] The Directive 2016/680 on the protection of natural persons with regard to the processing of personal data by competent authorities for the purposes of the prevention, investigation, detection or prosecution of criminal offenses or the execution of criminal penalties, establishes that: "Specific provisions of acts of the Union adopted in the field of judicial cooperation in criminal matters and police cooperation which were adopted prior to the date of the adoption of this Directive, regulating the processing of personal data between Member States or the access of designated authorities of Member States to information systems established pursuant to the Treaties, should remain unaffected, such as, for example, the specific provisions concerning the protection of personal data applied pursuant to Council Decision 2008/615/JHA", Par. 94.

[22] Report from The Commission to the European Parliament, the European Council and the Council, *Fifth progress report towards an effective and genuine Security Union*, COM/2017/0203 final.

decade later. The Commission has therefore used the enforcement powers it acquired under the Treaty of Lisbon in the Justice and Home Affairs area to launch infringement".

In this context, the Commission launched infringement procedures against Croatia, Greece, Ireland, Italy and Portugal, due to the failure to ensure the automated exchange of two out of three data categories and the lack of implementation of the European Decisions. This move by the Commission was aimed at encouraging those Member States to rapidly create national databases and to increase the efficiency of police cooperation in EU territory. Notwithstanding the Commission's intervention, this problematic non-compliance situation still persists today: considering the data provided by the Council of the EU Council in the Document 5197/1/20 Rev 1 of June 25th 2020,[23] infringement procedures against Italy and Greece are still open. In its study on the implementation and future of the "Prüm system", the Policy Department for Citizens' Rights and Constitutional Affairs of the European Parliament has underlined that "these delays may be attributed to various factors, primarily linked to financial and technical difficulties. For example, Greece, Italy and Ireland did not have DNA *database*s or dedicated legislation when the Prüm Decisions were adopted. Besides, these countries were severely hit by financial crisis".[24]

In addition to these considerations, influenced by the particular situation that still characterizes Italy and Greece and significantly affected the implementation process of the Prüm Decisions of other Member states, what should be carefully considered is the profound complexity characterizing the concrete implementation procedure of the data circulation and data exchange mechanisms.[25]

Even considering the Member States that have actually implemented the recalled Decisions, many differences can be identified, related to (a) the entry criteria for individual profiles;[26] (b) exclusion criteria of collected profiles;[27] (c) retention or destruction of biological samples;[28] (d) authorities asked to run and control the databases and officials responsible for safeguarding and protecting the retained data.[29]

[23] Council of the EU, *Implementation of the provisions on information exchange of the "Prüm Decisions"*, 5197/1/20 Rev 1, June 25th 2020.

[24] Policy Department for Citizens' Rights and Constitutional Affairs of the European Parliament, *Police information exchange*, cit., 19.

[25] See art. 36, Decision 2008/615/JHA.

[26] In some Countries (Austria), data of convicted individuals - without distinction regarding the seriousness of the crime - are included in the national databases, while in other cases only data of individuals convicted for a "serious" crime are included (Belgium, France).

[27] Profiles of convicted individuals can be stored in national databases for different amount of time (depending on the national legislation) after the conviction or even after the death of the convicted person (Finland). By contrast, data of individuals suspected of a crime but then not convicted or considered not guilty, are generally deleted from the database.

[28] "Biological samples" refer to biological liquids or tissues from which a DNA profile may be obtained. Some national provisions require the immediate destruction of biological samples right after the profiles have been obtained (Belgium), while in other Member states biological samples are subject to the same rules envisaged for DNA profiles.

[29] Generally, they are public officials within the law enforcement structure or under the guidance of the Ministry of Justice.

On the basis of these distinctions,[30] some macro patterns may be identified: one particular group of Member States (that includes, for example Austria and Finland) has adopted national legislations that allow for the collection and retention of a significant number of profiles and consequently ensures the possibility of exploiting and large amount of information for the hit/no hit operations. In doing so, those Member States demonstrated the tendency to enhance security to the detriment of citizens' privacy and data protection. Another group of States has instead opted for a higher level of protection of fundamental rights, determining restrictive criteria for the collection, entry and retention of genetic data in national databases; this choice demonstrated its limits, by negatively affecting the efficiency and effectiveness of the whole system (one example above all, Portugal). There are then some models that can be placed in between these two approaches, such as the French emergency-type one (affected by actual historical circumstances) or the German one which aims at developing a system characterised by a fine balance between the need for a high level of protection of citizens' rights and not creating conditions that are too stringent.[31]

An interesting legislative choice had been taken by Italy[32] where the national DNA database[33] was placed under the control of the Ministry of Internal Affairs, while a central laboratory responsible the sequencing and retention of biological samples was implemented within the structure of the Ministry of Justice. The decision to create two different structures of storage within two separate Ministries shows the intention to ensure a high level of expertise in each field and provide specific guarantees for the individuals engaged in the data collection: the operations of collection and matching of DNA profiles occur in a place physically separated from the place where the operations of extracting and retention of DNA samples are carried out.

The Italian legislator, despite the great delay, has achieved an appropriate balance between crime prevention and privacy and data protection, with particular regard to the delicate issue of data erasing. They have shown themselves to be aware of certain implications derived from the fundamental judgment of the Court of Strasbourg in the Marper case,[34] according to which an indiscriminate collection of genetic data, also for judicial purposes, constitutes a violation of art. 8 of

[30] F. SANTOS, H. MACHADO, S. SILVA, *Forensic DNA databases in European countries: is size linked to performance?*, in *Life Sciences, Society and Policy*, no. 9, 2013. For a more generic overview on this issue see L. SCAFFARDI, *Dati genetici e biometrici: nuove frontiere per le attività investigative*, in L. SCAFFARDI (a cura di), *I "profili" del diritto. Regole, rischi e opportunità nell'era digitale*, Torino, 2018, 37-64.

[31] For a thorough analysis of these models, in particular the ones adopted by Germany, UK and Portugal, see L. SCAFFARDI, *Giustizia genetica e tutela della persona*, cit., 69 ff.

[32] With the entry into force of Law no. 85 of June 30th 2009, published on the Official journal on 13th July 2009, Suppl. Ordinario n. 108, the national DNA database has been established. See on this point: G. GIOSTRA, *Gli importanti meriti e i molti limiti della nuova disciplina*, in G. CONSO, G. GIOSTRA (eds), *La disciplina del prelievo biologico coattivo alla luce della l. 30 giugno 2009, n. 85*, in *Giur. it.*, 2010, 1217 ff.; P. FELICIONI, *L'Italia aderisce al Trattato di Prüm: disciplinata l'acquisizione e l'utilizzazione probatoria dei profili genetici*, in *Dir. pen. proc., Speciale Banche dati*, 2, 2009, 6; L. SCAFFARDI, *Giustizia genetica e tutela della persona*, cit., 179 ff.

[33] After a long and complex legislative path, ended with the above mentioned Law no. 85/2009, the system became fully operative in 2018 after the adoption of the necessary implementing regulations and decrees during 2016 and 2017, that allowed the practical implementation of the national database.

[34] European Court of Human Rights, *S. and Marper v. United Kingdom decision*, [2008] ECHR 1581, 4th December 2008.

the ECHR.[35] In that case the Court was asked to evaluate the compliance of an English law, in force at that time,[36] to the fundamental rights protected by the ECHR: the Judges affirmed that the national provisions interfered with the right to privacy of the concerned individuals[37] due to the indiscriminate nature of the DNA samples' retention as well as the quantity and quality of personal information collected.

The Grand Chamber pointed out the undeniable utility of this forensic evidence (also including fingerprint conservation) and defined the conditions for proper use: the Court took the opportunity to reflect and make several useful observations related to the risks of stigmatization or indiscriminate use of forensic evidence.[38]

This important leading case brought about the revision of the English legislative framework in place at that time, but it also served as an important reference point for the development of similar legislation in other Member States.

4. The uncharted territory of Next Generation Prüm

The inefficiencies, delays and operative difficulties described in the previous paragraphs, together with the constant technological progress and the rise of new scientific techniques and investigative tools based on the use of personal data – genetic or biometric data for example – as well as the challenges determined by the spread of terrorism, organised crime and *cyber-crimes,* have prompted EU institutions, in particular the Council, to launch new initiatives by promoting studies, research and debates among experts and Member States 'authorities in order to modernise the Prüm Decisions, more than twelve years after their adoption. These intentions clearly emerge from the *Council*

[35] On the Marper case cfr. *ex plurimis*: A. SUTERWALLA, *DNA Discrimination*, in *New Law Journal*, Vol. 158, 2008, 505 ff.; A. JACKSON, *Public: Whose right is it anyway?*, in *New Law Journal*, Vol. 159, 2009, 187; C. NYDICK, *The British Invasion (of Privacy): DNA Databases in the United Kingdom and United States in the Wake of the* Marper *Case*, in *Emory International Law Review*, Vol. 23, 2010, 609 ff.

[36] In relation to *Section 64* of *Police and Criminal Evidence Act* of 1984, as modified in 2011 by the *Criminal Justice and Police Act*.

[37] The case was related to the claims lodged by two individuals: L.S., a minor arrested in 2001 and accused of attempted robbery and subsequently acquitted of charges, and Mr Marper, initially charged with sexual harassment on his wife, who then dropped the charges. Both subjects requested the destruction of their fingerprints and biological samples, previously collected and retained by the police during the investigations. After the rejection of their requests and having expired all internal remedies without success, they lodged their complaints before the European Court of Human Rights

[38] The Court underlined some considerations of the Nuffield Council on Bioethics, an independent body composed of physicians, jurists, philosophers, scientists and theologians who discuss about these delicate issues, similarly to the Italian Committee on Bioethics. See point 38: "According to a recent report by the Nuffield Council on Bioethics, the retention of fingerprints, DNA profiles and biological samples is generally more controversial than the taking of such bioinformation, and the retention of biological samples raises greater ethical concerns than digitised DNA profiles and fingerprints, given the differences in the level of information that could be revealed. The report referred in particular to the lack of satisfactory empirical evidence to justify the present practice of retaining indefinitely fingerprints, samples and DNA profiles from all those arrested for a recordable offense, irrespective of whether they were subsequently charged or convicted. The report voiced particular concerns at the policy of permanently retaining the bioinformation of minors, having regard to the requirements of the 1989 UN Convention on the Rights of the Child."

Conclusions on the implementation of the "PRÜM DECISIONS" ten years after their adoption (Conclusions 11227/18 18th July 2018): in order to reach the goals set by the Renewed Internal Security Strategy *2015-2020,*[39] the "effective and efficient use of the "Prüm Decisions" is considered essential for intensifying information exchange, cross-border law enforcement cooperation, for increasing mutual trust and for supporting solving of serious crime and conducting terrorist investigations" (p. 2).

On this basis, the Council requested that a possible amendment of the Prüm Decisions be considered, in order to extend the scope of their application and update the technical and legal requirements necessary for the functioning of the exchange system.

A wide and detailed discussion concerning possible modifications and forms of modernisation was opened by the *Working Party on Information Exchange and Data Protection* (hereinafter, DAPIX), [40] a preparatory body that, through the contribution of groups of experts,[41] started to evaluate the possibility of enhancing the so-called Next generation Prüm, keeping into consideration the necessity to guarantee the protection of personal data, as established in the GDPR and the Directive 2016/680.

The debate regarding possible modifications has moved along three different directions: a) innovate provisions governing data circulation and exchange in order to improve efficiency and effectiveness of the system; b) enhance the coordination between the Prüm system and other cooperation mechanisms for investigation and crime-fighting purposes; c) expand the categories of data included into the Prüm exchange mechanism.

A first important document about the potential developments of the Prüm mechanism is represented by the study conducted by Deloitte, upon request of the European Commission, concerning *the feasibility of improving information exchange under the Prüm Decision*, released in May 2020. In that document existing difficulties of the current system and possible feasible solutions have been considered with the goal of maximising efficacy of data circulation considering also the positive drive for new and more sophisticated technologies.

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[39] The so-called European Agenda on Security that defines the agenda, goals and priorities in this field, (COM(2015) 185 final. Strasbourg, 28.4.2015).

[40] The Council of the EU's official website explains that DAPIX "handles work relating to the implementation of legislation and policies on the information exchange and protection of personal data in the field of law enforcement. It also closely cooperates with Europol, especially regarding the Information Management Strategy (IMS) on streamlining cross-border information exchange. With respect to information exchange the working party is responsible for improving information exchange between law enforcement authorities of member states. With respect to data protection the working focuses on ensuring data exchange in compliance with current principles and rules on personal data protection", https://www.consilium.europa.eu/it/council-eu/preparatory-bodies/working-party-information-exchange-data-protection/.

[41] Four focus groups have been established, dedicated (respectively) to the evaluation of possible changes and improvements related to the exchange of: genetic data, fingerprints, data of vehicles' registration and finally – a relevant novelty – biometric data related to facial images deployed in systems of facial recognition. The four groups have been established with the aim of "setting out how to further develop the current information exchange mechanisms and to support the Commission's feasibility study on improving information exchange under the Prüm Decisions. The three groups focused on the existing data types already exchanged, whereas facial recognition was the subject of a fourth group established by the Council (Document 13356/19, 30 October 2019, not publicly available", Policy Department for Citizens' Rights and Constitutional Affairs of the European Parliament, *Police information exchange*, cit., 25.

Moving from the first of the mentioned areas, the envisaged solution is the introduction of new provisions on data exchange at EU level so as to make sharing and automated exchange easier and more efficient, in particular in the second phase of the *Prüm* mechanism, namely the step following the match between the data requested by a Member State and the one contained in the national database of another Country. As explained before, only in that case is the personal data of the subject linked to that genetic profile are actually shared. In such a delicate phase, different standards and procedures (e.g. some Member states require the intervention of a judicial authorities to authorise the submission of the data, while other prescribe supplementary controls) can ultimately represent an obstacle to the proper and efficient functioning of the exchange system.

For these reasons, enhancing automatic methods of communication of personal data may constitute a possible solution, in particular with regard to the exchange of fingerprints, considering the limited risk of false positive cases. The creation of a *central router*[42] goes in the same direction, aiming at receiving and sending all the requests coming from Member States' competent authorities with automated procedures: in this way the existing limits and problems related to the bilateral procedures of request and matching between two Member States could be overcome.[43]

Similarly, the EU institutions are now evaluating the possibility to modernize the existing Prüm system and to connect it with other cooperation or shared information systems among national law enforcement authorities: for example, cooperation with the European Search Portal (ESP)[44] or the

[42] On this point it is interesting to notice that this solution has been considered more appropriate than the creation of a "centralised information system, which has been rejected due to legal constraints on storing such data outside the national territory, for various reasons; processing personal data at EU level will be avoided; accurate statistical data at central level will be produced and technical difficulties posed by bilateral connections will be eliminated", Policy Department for Citizens' Rights and Constitutional Affairs of the European Parliament, *Police information exchange*, cit., 9.

[43] Another issue that has been underlined in this study is the inclusion, among the possibilities to activate and exploit the Prüm mechanism, of the purpose of searching missing persons: "new purposes will have to be added to the revised Prüm legal framework, so that searches with the aim of locating missing persons and identifying human bodies/remains could take place, even if no direct link to a criminal investigation exists". This obviously implies the necessity to define specific rules governing the guarantees for missing persons with regard to data protection "the fact that missing persons may include vulnerable groups of individuals, such as elderly persons, persons with mental health issues or children, should be taken into account. As a result, concerns are raised about the handling of data concerning missing persons in the same systems that process information on convicted criminals. Therefore, additional safeguards are required in relation to the retention of such data on missing persons", Policy Department for Citizens' Rights and Constitutional Affairs of the European Parliament, *Police information exchange*, cit., 27. Therefore it is evident that the political and legal debate on the issue is particularly delicate: the importance of guaranteeing a high level of protection of privacy and data protection rights (especially when referring to special categories of data like biometric and genetic data) along with the modernization and improvement of the Prüm system must be kept in mind.

[44] The European search portal has been established by art. 6 of Regulation (UE) n. 2019/817 of 20th May 2019 that establishes a framework for interoperability between EU information systems in the field of borders and visa. The portal aims at facilitating "the fast, seamless, efficient, systematic and controlled access of Member State authorities and Union agencies to the EU information systems, to Europol data and to the Interpol databases for the performance of their tasks and in accordance with their access rights and the objectives and purposes of the EES, VIS, ETIAS, Eurodac, SIS and ECRIS-TCN" (art. 6). As it can be read in the considering n. 10, "Interoperability between the EU information systems should allow those systems to supplement each other in order to facilitate the correct identification of persons, including unknown persons who are unable to identify

possibility for Europol and Interpol to have access to data exchanged among Member States or Third Countries according to the Prüm mechanism.[45]

Beside these innovations, a last area of intervention that should be carefully considered and evaluated is represented by the expansion of the data categories covered by the Prüm exchange mechanism, considering the huge impact that this decision could cause on the rights to privacy and data protection.

The study developed by the Commission has in fact taken into consideration the feasibility and utility of including, together with DNA, dactyloscopic data and data concerning vehicles registration, also facial images[46] – another biometric data alongside fingerprints – which represent the basis of advanced artificial intelligence technologies and cutting-edge systems of facial recognition.

Although these matters fall outside the scope of this paper, it is nonetheless essential to consider how the inclusion of these kind of data, particularly discussed among scholars[47] for their potential discriminatory outcome (also due to potential malfunctioning of algorithmic systems or possible still unexplored technical problems),[48] may result in a "*oversouveillance society*",[49] with significant

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themselves or unidentified human remains, contribute to combating identity fraud, improve and harmonise the data quality requirements of the respective EU information systems, facilitate the technical and operational implementation by Member States of EU information systems, strengthen the data security and data protection safeguards that govern the respective EU information systems, streamline access for the purposes of preventing, detecting or investigating terrorist offences or other serious criminal offences".

[45] A specific and complex question relates to the particular relationship with the UK: within the delays of the troubled Brexit process, specific agreements defining conditions, tools and procedures for data circulation – also comprised in the Prüm mechanism – have still to be concluded. This complex subject is intertwined with the provisions of the GDPR related to data sharing with Third Countries (status that will be acquired by the UK at the end of the Brexit process), which provide specific regulation and determine high standards and conditions to ensure that the data transfer is carried out according to a high level of data protection, even outside EU borders.

[46] "Given the maturity of the technology and its capability within the context of forensic law enforcement, recommends that the exchange of facial images be adopted in next Generation Prüm", Report conducted by Deloitte upon request of the European Commission (DELOITTE, *Study on the feasibility of improving information exchange under the Prüm Decisions*, 2020, 18).

[47] On this point see the considerations of the European Data Protection Supervisor in W. WIEWIOROWSK, *Ai and Facial Recognition: challenges and opportunities*, 21 February 2020, available at https://edps.europa.eu/press-publications/press-news/blog/ai-and-facial-recognition-challenges-and-opportunities_en. See also B. BUCKLEY, M. HUNTER, *Say cheese! Privacy and facial recognition*, in *Computer Law and Security Review*, 6, 2011; C. POPE, *Biometric Data Collection in an Unprotected World: Exploring the Need for Federal Legislation Protecting Biometric Data*, in *Journal of Law and Policy*, 2, 2018; R. KRISHAN, E. MOSTAFAVI, *Biometric technology: security and privacy concerns*, in *Journal of Internet Law*, July 2018.

[48] "The degree of accuracy in facial recognition technology is vital, so as to minimize the risk of false positive matches, namely results that may be unrelated to the investigation, or false negative results, when the facial recognition algorithm fails to identify correct matches. This is crucial since facial recognition technology will be used in the course of criminal investigations with the aim of identifying unknown perpetrators, therefore national authorities will perform searches on the basis of a facial image (a mug shot or a probe retrieved from a camera) against the full content of other national databases and the top results will be ranked. False positive matches in particular may have important consequences for individuals, who may be bothered by the police because of incorrect matching, be subject to criminal investigation and even be subject to discriminatory practices by national authorities", Policy Department For Citizens' Rights And Constitutional Affairs Of The

impacts on privacy and data protection of European citizens. The risk is to create a system of even more pervasive surveillance, along with a widespread genetic screening linked to the growing expansion of genetic databases.

All these considerations allow us to clearly understand how delicate the political and legal debate is around the expansion and modernisation of the existent Prüm system.

It becomes thus evident that it is necessary to deeply reflect, within the implementation of this new and important mechanism, on the implications and consequences on privacy and data protection rights, as well as on the principles of necessity and proportionality, that are meant to guide the processing of personal data, with particular attention to the so-called special categories of data, such as the genetic or biometric.[50]

The next steps towards the definition of new data exchange strategies should give a proper answer to the concerns and still open questions that characterize the extension of the Prüm mechanism.

Notwithstanding the direction that the reform of the Prüm mechanism will take in the coming years, the discussion around its modernisation shows the importance and the efficacy of the existing European data exchange system and at the same time reveals the inclination of the system itself to become what Wienroth has described as an *aspirational regime.* According to the author, "this regime provides context to the development and application of forensic genetic innovations by materially and discursively rationalizing and operationalizing research and technology uses at the transnational level. They are "aspirational" since their rationales and objectives are future-oriented: to develop technologies and techno-legal systems that can solve or prevent crimes, produce state security and public safety".[51]

Using this effective expression and extending its scope to all data collection and exchange systems of data, Toom and other authors affirmed that the *Next generation Prüm* project, while representing the evolutionary nature of such an exchange system, will also inevitably require "further investments in material infrastructures, including software packages, laboratory facilities, paperwork and (legal) rules and standards".[52]

In explaining such issues, it becomes clear that technological innovations and scientific progress in the data sharing field find a limit in their concrete implementation, which requires a huge effort by European and national legislators as well as law enforcement authorities.

European Parliament, *Police information exchange. The future developments regarding Prüm and the API Directive (Study requested by the LIBE Committee)*, September 2020, 19.

[49] The risk of an *oversurveillance society* through the use of genetic database has already been stressed by various authors, among which: P.E. TRACY, V. MORGAN, *Big Brother and His Science Kit: DNA Databases for 21st Century Crime Control?*, in *Journal of Criminal Law & Criminology*, 2, 2008, 635-690; R.E. RODRIGUES, *Big Bio-Brother is Here: Wanting, Taking and Keeping you DNA*, in *British & Irish Law, Educ. & Tech. Ass'n*, Vol. 2, 2007. Today the issue may become even more complex, following the potential implications deriving from the use of facial recognition systems.

[50] For a broader analysis on the provisions concerning these data within the GDPR framework, see R. DUCATO, *I dati biometrici*, in V. CUFFARO, R. D'ORAZIO, V. RICCIUTO (a cura di), *I dati personali nel diritto europeo*, Torino, 2019.

[51] M. WIENROTH, *Socio-technical disagreements as ethical fora*, in *BioSocieties*, 15, 2018, 1-18.

[52] V. TOOM, R. GRANJA, A. LUDWIG, *The Prüm Decisions as an aspirational regime: reviewing a decade of cross-border exchange and comparison of forensic DNA data*, in *Forensic science international: genetics*, 41, 2019, 54.

In the past, these challenges have already been able to restrain the expansion and effectiveness of the Prüm system, which faced delays in the development of databases and difficulties in the implementation of adequate and clear legislations, that were capable on the one hand to design solutions that guaranteed security and on the other hand could protect fundamental rights enshrined in the Nice Charter and in national Constitutions.

The legislator must find a proper equilibrium between the drive to exploit the potentialities of new technologies and the great quantities of data and, at the same time, respect fundamental rights which are today strongly safeguarded at European level, also through the case-law of the Luxembourg judges.[53]

[53] See for example on this subject the well-known case law of the CJEU on data retention for security purposes, starting with the judgment in Digital Rights Ireland (joined cases C-293/12 and C-594/12 of 8 April 2014) on the balance between the efficiency of the fight against serious crimes and the protection of privacy and data protection rights by the Luxembourg courts in this complex line of jurisprudence, ex multis O. POLLICINO, M. BASSINI, *La Corte di giustizia e una trama ormai nota: la sentenza* Tele2 Sverige *sulla conservazione dei dati di traffico per finalità di sicurezza e ordine pubblico*, in *Diritto penale contemporaneo*, 9 gennaio 2017; E. CELESTE, *The CJEU and the ban on bulk data retention: expansive potential and future scenarios*, in *European Constitutional Law Review*, 1, 2019. The EDU Court too has given a wide ruling on the massive monitoring and protection of fundamental rights, with relevant decisions, including recent ones, such as Centrum for Rattvisa c. Sweden and Big Brother Watch c. United Kingdom, currently referred to the Grand Chamber. See on this regards G. FORMICI, *La digital mass surveillance al vaglio della Corte Europea dei Diritti dell'Uomo: da Zakharov a Big Brother Watch*, in *Federalismi.it*, 23, 2020.

No sharp line between the natural and the synthetic: Biothetics and challenges to regulation

Mirko Đuković*

ABSTRACT: The code of human life stored in our cells is being extracted and transformed into biologically functional tissues at the same time it is also spawned with different synthetic materials that support those cells to mature and grow. Such products are essential of hybrid nature as they combine autologous and synthetic components, and thus making these products (that author labels as biothetics) difficult to categorize or put under existing know legal frameworks. This paper offers a brief review of the ethical, legal and social implications that regulators should be aware of. It also reflects on the bioconstitutional tensions that arise when novel technologies challenge the understanding of relations between our bodies, life and constitutions. By doing so, the paper examines the neoliberal underpinnings in the regulation of such a relationship.

KEYWORDS: Bioprinting; biothetics; regulation; bioconstitutionalism; ELSI

1. Introduction

According to the 2018 GODT (Global Observatory on Donation and Transplantation) report, 146 840 solid organs were transplanted. However, on average 20 people die daily, waiting for the donor. One of the solutions for the worldwide shortage of viable donors is bioengineering which aims at improving technologies to engineer cells, tissues and organs. One of the processes is the process of bioprinting. Using 3D printers in medicine might be one of the new technological thrilling opportunities for the humankind. The limited recourses in the matter allow us to set certain arguments that could be used for future debates and initial solutions to understand this technology and its implications on everyday life. Although printing entire organs are still not feasible at this juncture, bioprinting presents itself as a game-changing technology within the sphere of therapeutic medicine. Thus far scientist managed to print viable human tissues, such as skin grafts, cartilage and blood vessels have been used in therapeutic medicine with great success. The application of bioprinting extends beyond surgical wards, as pharmaceutical companies, cosmetics

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and personal care companies are as well interested in having this technology thrive. During COVID19 pandemic 3D printers were employed to alleviate shortages of personnel protective equipment, however, it was reported that 3D bioprinters are possible weapons against the pandemic. Bioprinting can help circumvent animal testing for covid19 vaccine as bioprinters could supply pharma with organoids that are viable for that same testing. The results in animals and humans differ and thus testing on tissues that mimic the human tissue would speed up the process.

This paper is based on my doctoral thesis research that is examining regulatory avenues for bioprinting technology. The research employs a comparative constitutional method in exploration of bioconstitutional and regulatory challenges in several jurisdictions, including the EU and the US. In the first part of the paper, I introduce the technology itself emphasizing the hybrid nature to it. As my thesis research indicates the term: "combination products" that regulators use in their narratives a bit redundant, and thus I offer a new terminology: biothetics. Further on I offer a short review of the possible ELSI read of the biothetics. The ELSI exploration consequently brings me to the proprietorship issues in bioprinting technology which comes from the fact that neoliberal "sacralization of property"[1] inevitably led to the profound desacralization of the human body on a very molecular level, as body as a commodity became a narrative with new biotechnological advancements. Taking interest in the relevance of what Jasanoff defined as bioconstitutionalism and her important work in understanding regulatory challenges in bioconstitutional legal order, I further explore how it was conditioned by the neoliberal regulatory narratives concluding that should the regulatory design remain unchanged for bioprinting, we might as well be sleepwalking into the dystopian future of Ishiguro, Orwell, or Crichton.

2. Revolutionizing biomedical engineering: bioprinting technology

Biomedical engineering is based on the processes that involve engineering principles and design concepts to medicine and biology for healthcare purposes, both diagnostic and therapeutic. In a matter of a few decades, this specific part of biotechnology emerged from experimental research programs to full flagship national public and private health institutes projects. When we are discussing bioprinting technology which is one of the biomedical engineering technologies, it is important to make a distinction between technologies such as tissue engineering, genetic engineering and neural engineering. Tissue engineering also referred to as regenerative medicine represents the creation of artificial organs from human cells using both synthetic and biological ingredients. Genetic engineering is a direct manipulation of the genes on a molecular level to transform or alter the structure of the targeted genes. And finally, neural engineering is a discipline that focuses on the repair, replacement or enhancement of the neural systems using engineering techniques. In general, the most recent developments in the bio-fabrication processes provides for manufacturing of the complex tissues constructs with a higher degree of biomimicry to native tissues.[2]

[1] T. PIKETTY, *Capital and Ideology*, Cambridge MA, 2020, 122.

[2] L. MORONI et al., *Biofabrication: A Guide to Technology and Terminology*, *Trends in Biotechnology,* 36, 4, 2018, 384-402.

In a word, all these technologies or processes represent an ongoing evolution that is happening in the bioengineering and there is not much we can do but to wait for the revolutionary outcomes or we could anticipate the outcomes based on the few decades-long debates and prepare ourselves to keep up with science. One thing is certain: once the results of these technologies are out at the market we have to mobilize our entire experience based on the decades of dealing with technological improvements so that we can accommodate the technology that can fit the common human values without deriving us from our guaranteed rights and freedoms. The perils of some of these revolutionary technologies are already being reported to the society as biohackers managed to by using CRISPR change their DNA, and at the moment we cannot stop it.[3] On the other hand, the tissues engineering had its major success when Anthony Atala with his team managed to engineer human bladder tissues for "patients with end-stage bladder disease by isolating autologous bladder urothelial and muscle cells, expanding the cells in vitro, and attaching them to biodegradable three-dimensional matrices".[4]

The origins of the groundbreaking science behind bioprinting of human tissues are in the brilliant realization that additive manufacturing and bioengineering can be combined. Additive manufacturing, rapid prototyping, stereolithography or most commonly 3D printing is a process that turns blueprints into solid physical objects and this technology was in its nascent stage in early 1980ies. Applying such logic first two-dimensional structures made of cell adhesion proteins were developed by Robert J. Klebe in 1988. In 2002, Japanese scientist Makoto Nakamura realized that size of cells corresponds to the size of a drop of ink and his experiment showed that cells could survive the printing process.[5] But the first modification of the 3D printer into the ink-jet printer to print scaffolds was done by Thomas Boland and his team in 2003.[6] A year later Gabor Forgacs discovered that mammalian cells have physical mechanisms to form multicellular aggregates of a controlled shape and size and he managed to build the prototype of a printer that enables precise placement of cells that could form a tubular structure with a tissue as a final product.[7] The patent of Thomas Boland was published in 2006 and in 2009 the company "Organovo" created the first bioprinter based on these technologies.

So what is bioprinting? Simply put it is the use of the 3D printers to print human tissues layer by layer by fusing the living human cells into the scaffolds in a controlled environment. Most often it is described as one of the biofabrication technologies that can overcome the difficulties of producing the exact mimic of the native tissues since authors agree that by placing multiple types of cells in a

[3] F. FORMAN, [Tt] NS 3152: *Biohackers Are Using CRISPR on Their DNA and We Can't Stop It*, 25 November 2017, http://postbiota.org/pipermail/tt/2017-November/020889.html, last visited 15.12.2020; A. PEARLMAN, *Biohackers Are Using CRISPR on Their DNA and We Can't Stop It,* in *New Scientist* <https://www.newscientist.com/article/mg23631520-100-biohackers-are-using-crispr-on-their-dna-and-we-cant-stop-it/, last visited 15.12.2020.

[4] A. ATALA et al., *Tissue-Engineered Autologous Bladders for Patients Needing Cystoplasty*, in *Lancet,* 367, 1241, 2006, 1245.

[5] M. NAKAMURA et al*., Biocompatible Inkjet Printing Technique for Designed Seeding of Individual Living Cells* 11, 2005, *Tissue Engineering* 1658.

[6] T. BOLAND, W.C. WILSON, T. XU, *Ink-Jet Printing of Viable Cells*, 31.

[7] 'Scientific Origins' (*Organovo*), https://organovo.com/science-technology/bioprinted-human-tissue/scientific-origins/, last visited 15.12.2020.

designed matrix could bring us closer to producing more complex human tissues.[8] Bioprinting is a process that requires extraction of the human cells (ideally patient's cells), or reprogramming of the embryonic stem cells which are replicated to produce ink that is mixed with a hydrogel that serves as a living environment for the cells to bind and survive, a bioprinter and CAD images of the desired tissue to be printed. For cells to bind and mimic the natural processes they need a synthetic scaffold in which they communicate, exchange nutrients and multiply to form a tissue. Thus far, relatively simple tissues such as cartilage, skin grafts, vascular tissues, lung tissues and bones are being printed and applied in treatment. More complicated tissues and organs such as heart, liver or kidney, meaning those that are combined of several different cell types are more challenging to be printed, but miniature versions of them are being printed, thus making bioprinting promising technology in saving those who wait on transplant lists, living in hope that the organ they need will find its way to them.[9]

2.1. Biothetics

As it was mentioned, the scaffolds mimic the extracellular environment so that cells that are placed into them through bioink nozzles can live and form tissues. Here is what especially draws my attention, the fact that the process of printing a tissue requires synthetic materials, which essentially makes products of bioprinting non 100% human. Some authors question if because of synthetic scaffolds that are needed for the maturation of the tissue, these products might be patentable as they are non-natural occurring.[10] Here, we can observe the tension between non-organic and organic, or synthetic and natural components of bioprinting processes and products. We witness how techno-medical advances are challenging the very definition of life: while the biological definition of life focuses on biological and physical processes the legal focus on life entitlements or consequences of such advances.

The biotechnological progress that marked last few decades are groundbreaking moments of discovering possibilities of human cloning, *in vitro* productivity of human tissues, human stem cell research, genome editing, development of pluripotent stem cells, etc. Yet, these biotechnological advancements constantly challenge existing legal and ethical frameworks. For example, pluripotent stem cells have the ability to evolve into any type of cells and could play important role in bioprinting of human tissues. Research on stem cells is of great significance not only in medicine but also in physiology, medical education, biology and pharmaceutical industry. This comes from the fact that stem cells have unique abilities in proliferation and differentiation in comparison to another type of cells. Research indicates that stem cells "serve as a reliable cell source in bioprinting for tissue engineering, regenerative medicine, drug testing and cancer studies".[11] However, the production of

[8] Z. XIA et al., *Tissue and Organ 3D Bioprinting*, in *SLAS TECHNOLOGY: Translating Life Sciences Innovation*, 2018, 1.

[9] N. NOOR et al., *3D Printing of Personalized Thick and Perfusable Cardiac Patches and Hearts*, in *Advanced Science,* 6 2019.

[10] R. JACOBSON, *3-D Bioprinting: Not Allowed or NOTA Allowed?*, in *Chicago-Kent Law Review,* 91, 2016, 1129.

[11] S. DING et al., *Bioprinting of Stem Cells: Interplay of Bioprinting Process, Bioinks, and Stem Cell Properties*, *Acs Biomaterials Science & Engineering,* 4, 2018*,* 3108.

stem cells confronts the protection of unborn life and human dignity. While the EU laws prohibit patentability of inventions based on the embryonic stem cells from embryos *in vitro*[12], the UK is more flexible and under US laws Federal Government funding is not available for research that entails the destruction of the human embryo.[13]

Even with different types of bioprinting technologies, be it due to the cell types that could be used in bioprinting process or the kind of bioprinting process applied, the synthetic component plays important role in tissues production which essentially makes bioprinting a hybrid kind of technology with a dual nature to it. In some of the guidelines and analysis of the technology offered by regulators, these products are often called "combination products", and even so it can be contested that it is not clear what that means for bioprinting, as "combination products" are those that combine drug, a device and biologic product. In the EU, however, the legal framework recognizes medical devices and medicinal products. In my view, this seems misleading, and linguistically redundant. Because of this, I introduce the term "biothetics" as a term that, in social sciences, should be used to describe products that are the result of *in vivo* and *in vitro* processes. Biothetics are also products based of autologous material (materials that derive from the human body) and non-biological materials, such as synthetic materials (such as biodegradable polymers, hydrogels and dopamine-modified alginate and polydopamine hydrogels) to keep the autologous materials alive.

The word biothetics in my research refers to the processes and products of bioprinting. The word is comprised of three words which come from Indo-European language family: biology (*noun*, Greek βίος; Romanized: bíos meaning "life") and thesis (*verb*, New Testament Greek τιθημι; Romanized: tithēmi meaning "to make, to place, or to establish") and synthétique [*adjective,* late 17th century French or modern Latin syntheticus, from Greek συνθετικός (synthetikos), based on συντίθημι (suntíthēmi) "to place together"] meaning made by chemical synthesis, especially to imitate a natural product. The adjective is biothetic, the verbal noun being biothesis, and the noun that describes the products would be biothetics.

3. Preliminary considerations of some ethical, legal and social implications of biothetics

Bioprinting falls in the category of 3D printing but the laws that govern 3D printing technology are not entirely applicable to the bioprinting, as obviously the application of the technologies is very different. The 3D printing is already having an immense impact on the different aspects of modern living, but bioprinting is about to revolutionize the medicine and for a few years now, authors are examining what are some of the most pressing ethical, legal and social implications of this technology.

[12] *Oliver Brüstle v Greenpeace eV C-34/10* (Court of Justice of European Union). More on this also in: Lucchi

[13] The Dickey-Wicker Amendment is the name of an appropriation bill which prohibits the Department of Health and Human Services (HHS) from using appropriated funds for the creation of human embryos for research purposes or for research in which human embryos are destroyed. See also: *Sherley v. Sebelius* (2010).

3.1. (Bio)Ethical implications

Recent incorporations of bioethical norms in the human rights legal framework that made sure that science is "no longer regarded as the value-free domain of research".[14] This normative approach to bioethics research according to Sandor is distinguished in the two major lines of thought: the one exploring the boundaries of life (embryo research, assisted reproduction, etc.) and one exploring the frontiers of the human body (human tissue research, human DNA research, etc.).[15] In that respect, there are a few categories that caught attention when it comes to bioprinting. The first considers the ethics related to the clinical translation which again presupposes the discussion on pre-clinical and clinical testing. Technologies such as this one have irreversible impacts on biological and physiological settings of our bodies. Having hybrid object implanted in our body requires knowing exact reactions and consequences it will have on us, as presumably, we get to live with whatever comes in the post-clinical phase. For the time being, testing on animals, with all the ethical contestations this has, is the only way scientists can know what are the potential reactions to it as computer modelling or in vitro studies cannot fully answer all the concerns. This pre-clinical testing shall be followed by clinical testing where the new ethical dimension arises as the testing is done on selected individuals. The *in vivo* studies are required to show the long-term results of the implanted tissues, as well as to explore possible side effects that might emerge. The computer testing and animal testing cannot show with 100% efficacy of how the implants will behave in humans. The second category of ethical implications of biothetics is the one briefly voiced before, and it concerns the use of stem cells, especially if bioinks are composed of replicated embryonic stem cells. There are disagreements amongst regulators, ethicists and lawyers on this matter as some of them list at least ten ethical concerns in the application of embryonic stem cells.[16] Besides, the issues of using xenogeneic cells that derive from other species of animals and cells cultivated via chimera technology also rise questioning eyebrows. This especially when we take into consideration the biological risks of pathogens and similar agents being involved or the reservations that are rooted in various religious views in respect to such cells. The last argument usually comes hand in hand with the culturally and socially shaped view that such technologies imply that "humans are playing God" and that this to a certain extent reminds us of cloning. The third category is the one that is again connected to the source of the cells being used to produce bioinks, but its objective is towards the donation of cells. This category covers questions concerning the privacy of the donor, the informed consent, the invasiveness of the cell obtaining process as well as the issues around ownership of the biothetics.[17]

[14] J. SANDOR, *Bioethics and Basic Rights: Persons, Humans and Boundaries of Life*, in M. ROSENFELD, A. SAJO (eds), *The Oxford Handbook of Comparative Constitutional Law*, Oxford, 2012, 1143.
[15] *Ibidem.*
[16] R. DE VRIES et al., *Ethical Aspects of Tissue Engineering: A Review*, in *Tissue Engineering, Part B: Reviews,* 14, 2018*,* 367.
[17] P.P. VAN DEN BERG et al., *Ethical Issues Regarding the Donation and Source of Cells for Tissue Engineering: A European Focus Group Study*, in *Tissue Engineering*, 17, 2011, Part B: Reviews 229, fig 2.

ISSN 2284-4503

3.2. The legal implications

Some authors such as Tran suggest that bioprinting brings more benefits that outweigh the risks in comparison to the other biotechnologies.[18] However, there are many issues to be resolved before we face the first entirely printed organ to be transplanted into a living human. Besides the blurring of boundaries between autologous material and synthetic materials, there are more predominantly regulatory aspects that need to be explored, such as ownership of data, intellectual property and privacy; issues of safety and informed consent; and standards and requirements for product development, validation and testing meaning quality assurance, preservation of genetic data. Also, biothetics are subject of many transactional interests between various actors and intermediaries thus regulators should be aware of the role of the private investors and their role in the regulation and distribution including health care organizations, patient rights organizations and insurance groups. The policymakers ought to set up regulatory panels[19] comprised of relevant authorities, scientists, bioethicists, academics, lawyers who would follow the development of the technology and be ready to answer challenges that arise. Such challenges might not be in sight at the moment, but as we have seen with other similar technologies, legal frameworks are constantly in a struggle to keep up with science. Hypothetically one of the challenges might be if bioprinting is used not only to save someone's life but also to enhance the features of the printed organ. Introducing such technology to those who would abuse the potential of it would greatly impact existing inequality in access to health care. Another potential abuse is using this technology to enhance someone's ability to compete in sports, which could happen when the line between treatment or prevention and enhancement is blurred. Can you imagine a tennis player competing in his fifties as if he was in his thirties, as he has means to support such enhancement? I surely can.

The before mentioned ownership of biothetics is the one that invites both legal and philosophical discussion: who owns our bodies and body parts? Under the principles of constitutional law, property rights protect the autonomy and privacy as fundamental rights in different ways, from country to country, but in general, in terms of liberal democratic constitutions, they give a right to any individual to make a personal decision free from state or regulation.

According to Jasanoff, the significant milestones in the life sciences and technologies constitute a bioconstitutional rather than constitutional relationship between states and citizens.[20] It brings about "far-reaching re-orderings in our imagination of the state's life-preserving and life-enhancing functions – in effect, a repositioning of human bodies and selves in relation to the state's legal, political and moral apparatus".[21] To this, I would add Sandor's observation that even though bioethics and human rights are distinctly different disciplines there is no established method to recognize when some universal norms became basic rights in the field of bioethics. Thus, what Jasanoff explores through the constitutional significance of scientific breakthroughs that have an impact over our lives, where our bodies are nexus of law and life, are what Sandor identifies as the

[18] J.L. TRAN, *To Bioprint or Not to Bioprint*, in *North Carolina Journal of Law & Technology,* 17, 2015, 178.

[19] The European Parliament has already made inquiries and has published several in-depth analysis of the technology.

[20] S. JASANOFF, *Reframing Rights: Bioconstitutionalism in the Genetic Age,* Cambridge MA, 2011, 4.

[21] *Ibidem.*

animating force behind international and supranational organizations attempt to incorporate bioethical norms into an international binding and non-binding legal framework.

The legal or better said in this context bioconstitutional and philosophical dilemma if we own our bodies is not new and it is not brought by technology subjected to this paper, but it again contests some of the existing narratives in the field. As it was mentioned earlier biothetics are hybrid products that have dual nature to it, and this fact might pose new contestations in the domain of property in body. Observed through Jasanoff's understanding of bioconstitutionalism and Sandor's perception of correlation between normative bioethics and human rights, I claim that biothetics are that major technological breakthrough that could shift the paradigm around personhood, autonomy, right to privacy, self-ownership or even ownership interests as defined in the *Moore case*. Supreme Court of California ruled that individuals do not have ownership interests in their cells once removed from their bodies, but recognized the importance of informed consent as bioethical principle *sine qua non*. Brownsword challenges such logics as he rightly wonders how is it possible that someone can have property rights over my body or me over someone else's body all the while none of us has property rights over our own bodies?[22] The paradigm of self-ownership is changed on the premise of the bioconstitutional shifts that come with new technologies and scientific advancements which changes the relation between us and the state, but also clearly our interests over our bodies and those of science.

So, the question is what is the capacity of human tissues and bodily materials once removed from the body? If we consult Locke today, he might have told us that "every man has a property in his own person".[23] However, applying his labour-desert theory, we can argue that we do not own our bodies as we did not labour to produce them but we do own the moral agency and the bodily tissues that we produce (for example, producing ova that are used in stem cells technology). Kant on the other hand would deduct that it would be morally wrong to "reduce subjects to objects in some essential sense" and thus we would not have the agency to sell our biothetics. In her comprehensive review on human body and property, Rao contests that once society permits alienation of the body part, that body part constitutes property no matter the legal status. However, she explains: "if body parts are deemed inalienable and unassailable, they should be regarded as the subjects of privacy interests rather than the objects of property law".[24]

Jasanoff draws a line between the issue of property and personhood, as the first one is within the dimension of intellectual property rights as the notion of what is property evolves once technology creates entities "between living and nonliving, human and nonhuman".[25] Which again confirms that biothetics as hybrid entities can challenge conceptions of property rights within the novel bioconstitutional capacities of state-citizens relationship.

[22] R. BROWNSWORD, *Rights, Regulation, and the Technological Revolution,* New York, 2011, 61–62.
[23] J. LOCKE, *Two Treatises of Government*, e-book version, 1980, 45.
[24] R. RAO, *Property, Privacy, and the Human Body*, in *Boston University Law Review,* 80, 360, 2000, 459.
[25] S. JASANOFF, *The Ethics of Invention: Technology and the Human Future*, New York, 2016, 189.

ISSN 2284-4503

3.3. Social implications

The previous discussion leads us to the short review of what are some of the social implications of the bioprinting technology. The last couple of years media suggested that in the future humankind will be blessed by the possibility to biofabrication personalized organs and thus the global shortage of viable organs for transplantation will be diminished.[26] Some authors argue that this technology has the same revolutionary and democratizing effect as book printing had in its time in their applicability to regenerative medicine and industry.[27] In addition, there are currently no regulations on bioprinting as well there has been no litigation on bioprinting. Some patent applications were made, but this is a topic of entirely other research.

As history teaches us, every time a new technology is introduced to the people there are two imminent outcomes. The first one is the overall social hype about the new technology and its reception in society. The second one is that as soon as the technology unveils its power and potential to the users, it could end up into the hands of people who did not have such power before. As one technology emerges something illicit happens in its early stage,[28] so it takes some time to learn how to regulate it and catch up with it. Discussing the "tinkering with humans" Jasanoff observes that "manipulations of human biology raise even more complex issues for ethics, law, and policy [...] in biomedicine, the greatest fear centres on violating human integrity and eroding the fundamental meaning of being human".[29] This inevitably leads to the question if bioprinting will become a human/genetic enhancement tool as new biotechnologies are forcing us to re-think and re-evaluate in what kind of world we want to live. Finally, it also points that biothetics have a potential to raise the inequality in access to necessary therapies, and thus instead of helping population it might help only certain categories of people, meaning those who could afford such treatments. It will be extremely difficult in the current free-market setting to safeguard the innovation which in essence should be assessable and affordable for public health. Observed in the frame of distributive justice argument, new technologies can only advance justice when it will benefit marginalised and poor. On the other hand, the instrumentalization of marginalised groups comes at play as well in the context of the black market of bioprinted organs potential. The tissue transfer follows the rules of power and wealth, meaning the most vulnerable class of society that is prone to sell their bodily products. Besides the fact that organ black markets are thriving in the war-affected zones, trading with gametes and renting out wombs for surrogacy are no exception.

In *The gift relationship* Titmuss compared the national blood donation systems in the UK and the US. The policies around blood management grew more complicated over time as in the US unlike in the UK government allowed for private supply chains. The basic premise of private blood banks was that people were more likely to sell their blood rather than volunteer to donate. He observed that for-profit blood banks exploited the regulatory gaps and were able to compensate the problems in

[26] J. GALLAGHER, *Doctors 3D-Print "living" Body Parts'*, in *BBC News*, 16 February 2016, https://www.bbc.com/news/health-35581454> last visited 10.01.2021.

[27] N. VERMEULEN et al., *3D Bioprint Me: A Socioethical View of Bioprinting Human Organs and Tissues*, in *Journal of Medical Ethics*, 2, 2017, 1–7.

[28] K.S. KRUSZELNICKI, *Mouse with Human Ear*, https://ab.co/39QpNFL, last visited 22.01.2021.

[29] S. JASANOFF, *The Ethics of Invention: Technology and the Human Future,* 120.

supply that hospitals were facing, which essentially turned blood into a commodity that was predominantly procured from indigent and homeless.[30] The gift system that was established prior to this was something that kept nations resilient and blood donation was an altruistic and valuable part of the community culture. For-profit blood banks were a mere representation of what neoliberal market rationalism can do to healthcare and thus he was a strong advocate against such a system. Not only that the blood as a commodity was devalued of its ontological quality it was indicating a deep social disparity that affected the very aim of distributive justice. Should bioprinting be left to the market regulation narratives, it can be presumed that similar occurrences as described by Titmuss would take place.

In the section where I introduced some of the ethical issues around the bioprinting, it is inescapable to make the connection with social issues and religious views that will influence the overall perception of the technology. There are alternative ways to using embryonic stem cells but studies indicate that embryonic stem cells have the greatest potential to be reprogrammed and endure printing processes.[31] The general public in different jurisdictions can show discontent towards technology that is developed around the destruction of embryos. Similarly, certain countries, even belonging to the same legal space explicitly prohibit therapeutic cell cloning (Spain, Sweden, Austria, Germany) and some do not (Denmark, the Netherlands). This perfect example is the EU, where the issues of public bioethics and ethical standards of new technologies are conferred to the competence of member states due to the constitutional principle of subsidiarity, although Charter of Fundamental Rights of the EU offers a catalogue of common bioethical principles.[32]

It is important to acknowledge that the benefits of bioprinting are many. It would potentially stop animal testing in all the relevant industries, it would undoubtedly change the future for transplantation surgery and hopefully would terminate immense financial costs. In terms of education and studying of deceases it would help science to re-evaluate the courses of treatments, it would avoid so-called “yuck” factor as it does not instigate reactions such as the ones people usually have when hearing about chimera technology or xenotransplantation.[33]

4. Current legal frameworks: comparison

To this day I did not learn of any country in the world specifically regulating this technology, and in my research, I mostly focus on the EU and US, and thus I will make a brief review of the current state of play.

In July 2018, the European Parliament published an in-depth analysis of *Additive bio-manufacturing: 3D printing for medical recovery and human enhancement*. According to this analysis, the main challenge for the regulators will be defining and categorizing the processes and products of the technology. It recognizes the difficulty to subsume biothetics in the existing legislative framework. For example, In Vitro Diagnostic Device Regulation 2017/746, does not provide any particular

[30] R.M Titmuss, *The Gift Relationship: From Human Blood to Social Policy*, New York, 1971, 119.
[31] S. Ding et al., *op. cit.* 11.
[32] J. Sandor, *Bioethics and Basic Rights: Persons, Humans and Boundaries of Life*, 1146.
[33] N. Brown, *Xenotransplantation: Normalizing Disgust*, 8 *Science as Culture,* 1999, 327.

ISSN 2284-4503

guidance on these distinctions and its provisions do not state clearly how substances of human origin should be judged.[34] Similarly to the observation, I offered in the previous section, this difficulty comes from the fact that this technology is of a dual nature. It remains unclear if biothetics, referred to as "combination products" are biological product or if a non-biological component to it makes it a medical device or if it falls under the advanced therapy category.

To answer that question, the so-called "Mode of Action (MoA)" determines in which way the finished product will be classified, and it depends on the achieved purpose of the product. As an example authors apply these logics on the nasal implant which "consists of substantially manipulated chondrocyte (cartilage) cells, is presented as having properties for regeneration and repair of a human tissue, and contains (as an integral part) scaffold material that fulfils its function as a medical device when deployed in the patient, the product falls within the definition of a tissue-engineered combined ATMP, as defined in article 2(1)(d) of Regulation (EC) No 1394/2007/EC [15]". This however is again contested by the very notion that biothetics represent the unclear definition of what they represent, or they can have a different intended purpose in different patients. This is well acknowledged in the Report, as it suggests that "the manufacturer may not have sufficient data and/or scientific knowledge in the early development phase to identify the principal MoA and thereby the candidate ATMP classification".[35] European Parliament analysis indicates that even recently adopted Medical Device Regulation (MDR) 2017/745 does not regulate bioprinting, and the provisions of the Regulations are not addressing the issue that products of bioprinting are custom made and not large scale industrially produced, and thus it becomes difficult to ensure the quality of the "in-house custom-made" biothetics. The entire premise of the technology is printing human organs on demand for a specific patient, the current legislative framework is challenged by it.

When it comes to the US, in 2016 the FDA issued draft guidance on the Technical Considerations for Additive Manufactured Devices which had the goal to obtain public feedback. It is organized in the two areas, the design and manufacturing considerations and device testing considerations. It recognized that bioprinting has versatile application in terms of medical devices, biologics and drugs.[36] Just like it was the case in the European Parliament report, the preliminary guidance issued by the FDA does not address the classification of the products with respect to its composition. Under 21 CFR Part 1271.3(d) provision one can argue that biothetics are covered, to a certain extent as this provision states that: "Human cells, tissues, or cellular or tissue-based products (HCT/Ps) means articles containing or consisting of human cells or tissues that are intended for implantation, transplantation, infusion, or transfer into a human recipient. Products such as bone, ligament, skin, dura mater, heart valve, cornea, hematopoietic stem/progenitor cells derived from peripheral and cord blood, manipulated autologous chondrocytes, epithelial cells on a synthetic matrix, and semen or other reproductive tissue". Furthermore according to the 351(a) of the 42 USC 262: Regulation of biological products, the biological products are those that are "applicable to the prevention,

[34] A. FERRARI et al., *Additive Bio-Manufacturing: 3D Printing for Medical Recovery and Human Enhancement (European Parliament's Science and Technology Options Assessment STOA) 2018,* 89.

[35] *Ivi,* 90.

[36] Center for Devices and Radiological Health, *'FDA's Role in 3D Printing'*, 2019, https://www.fda.gov/medical-devices/3d-printing-medical-devices/fdas-role-3d-printing, last visited 10.01.2021.

treatment, or cure of a disease or condition of human beings" and under 21 CFR 3.2(e) that includes "a product comprised of two or more regulated components, i.e., drug/device, biologic/device, drug/biologic, or drug/device/biologic, that are physically, chemically, or otherwise combined or mixed and produced as a single entity". This short comparison indicates that both jurisdictions lack proper definitions and regulatory framework which would offer a unison and consistent protection for all the relevant stakeholders.

The EU pays great attention to the IP and data protection aspects. According to the analysis, it is not clear if the current IP framework can sufficiently protect both files and those using the technology for non-commercial purpose. As indicated before, due to the nature of biothetics is it safe to claim that they are not patentable due to the human organism exception rule. And finally, if the applicability of the morality clause will be extended to biothetics, especially when they are made of embryonic stem cells which are protected due to the understanding that human dignity extends to the embryos.[37] In the domain of the IP related discussions, it is worth realizing that bioprinting brings the question if it should be categorized as machines used for a medical purpose which would make the technology patentable or it is a medical technique that involves in vitro and in vivo processes and thus is non-patentable.[38] For other authors, it is an issue of ownership and the value of the products for different parties in the process.[39] Li warns about dangers from the monopoly that stem from the contemporary ideology to protect scientific innovations and due to the potential that this technology has to save human lives, she advocates so-called "portfolio approach" to licensing as it would be a "balanced pathway to 3D bioprinting could be built to avoid overreliance on the patent system".[40]

Further on GDPR extends to the consent, and thus any processing of patient's data must meet GDRP criteria. However, nowhere does GDPR provide clear guidance on how to treat innovation such as bioprinting. The consent to remove, store, process and use both data and tissues must be such to minimise the risk of any harm. However, there is another layer of data protection that I would like to discuss here: there is an additional value to creating data repositories for scientific research. Researchers build those repositories to improve medical practices as well as medical education, to observe and map important points in human evolution but they can also help them understand how a certain person lived, died etc. Even when the consent is obtained it is hard to make sure that data stored in the scientific community will remain protected from illegal downloads. The ethical standards behind the data protection also differ from country to country, and we should not be so naïve to think there could be no malicious attempts in examining patient's data from other countries and continents. One example, in particular, comes to mind: the so-called "race science" which aims to prove that humans can be divided into race categories based on their specific biological and physio-anatomical features. It should be noted that even in the domain of transporting and exchanging human biological materials, countries have different regulations which rarely regulate the protection of digital data on human tissues. As I previously mentioned human bioprinting

[37] *Oliver Brüstle v Greenpeace e.V C-34/10* (n 12).

[38] P.H. Li, *3D Bioprinting Technologies: Patents, Innovation and Access*, in *Law, Innovation and Technology,* 6, 2014, 282.

[39] J.T. Harbaugh, *Do You Own Your 3D Bioprinted Body? Analyzing Property Issues at the Intersection of Digital Information and Biology*, in *American Journal of Law & Medicine*, 41, 2015, 167.

[40] P.H. Li, *op.cit.* 301.

hardware is not difficult to obtain, downloading data on human tissues is a matter of seconds, protection from both is a matter of years. This is mostly because it is a well-established notion that data and source materials (cells and tissues stored in biobanks) are not comparable and are distinct. Consequently, not only that science is constantly slipping away from regulation, but also the regulatory models need an upgrade.

5. Neoliberal underpinnings, regulatory models and bioprinting

Several international, regional and supranational documents, binding and non-binding, have been introduced to protect health, environment and society from the risks of the potential uncertainties that any scientific discovery could have. In that sense, most of the documents are centred around human dignity as an ultimate focal point which could guarantee the protection of rights. Post II WW the human rights norms in bioethics were a response to violations of basic ethical norms in all sorts of research on the human body, to the molecular level but today the focus shifted to the possibility of transformation of human nature.[41] Yet, some believe there is no need to adopt new legal norms. In my view, this is not a question if we need to adopt new legal norms but in what way can we re-imagine regulatory designs so that we can identify what rights we ascribe to biothetics. It would be premature to claim that bioprinting brings novel rights, but as this paper suggests, it is challenging many aspects of law and regulation.

What was set by Ulrich Beck as the thesis of "reflexive modernization" Jasanoff further elaborates through the thesis that although "scientific and technical advances bring unquestioned benefits they also generate new uncertainties and failures, with the result that doubt continually undermines knowledge, and unforeseen consequences confound faith in the progress".[42] Recognizing not only the advances in technology but also the uncertainties, failures and these unforeseen consequences, Brownsword points out when regulating it is the essential need to frame our inquiries as there are no doubts to why biomedical technologies should be regulated, however, the relationship between law and regulation is unclear.[43] He inquires whether a law is to be understood as a broader enterprise than regulation or narrower? Relying on work of Julia Black he posits that regulation is primarily about channelling behaviour so legislation is a species of regulation.[44] Meaning, regulation is a broader concept. But this comes with a warning because regulation is a "constant learning process" and that is especially important in the case of the emerging technologies with uncertain risks and knowing *when, what and how* to regulate is a challenge itself.[45]

Having said that, governments have developed so-called predictive methods to assess risk management, cost-benefit analysis etc. and the whole purpose is to facilitate management and

[41] J. SÁNDOR, *The Challenge of Genetics: Human Rights on the Molecular Level,* in A. VON ARNAULD, K. VON DER DECKEN, M. SUSI (eds), *The Cambridge Handbook of New Human Rights: Recognition, Novelty, Rhetoric*, Cambridge, 2020, 356.

[42] U. BECK, *Risk Society: Towards a New Modernity*, 1992; S. JASANOFF, *Technologies of Humility: Citizen Participation in Governing Science*, in *Minerva,* 42, 2003, 224.

[43] R. BROWNSWORD, *op.cit.* 6.

[44] *Ibidem.*

[45] *Ivi,* 123.

control even in areas of high uncertainty.[46] Jasanoff analyzes these tools in-depth and highlights their three major flaws or limitations. She points out that the first limitation is in the fact that in making any assessments, this tool always focuses on the known and omits to make any other predictions. Besides, when applying predictive analysis agencies involve experts and exclude political discussion so the objectivity of the analysis is blurred as it is carried out away from the general public. And lastly, these tools are unable to "internalize challenges that arise outside their framing assumptions."[47] Meaning that unknown features of the technology are left out from the analysis and are nowhere to be found in legislative processes.[48] What Jasanoff suggests is that predictive methods need "technologies of humility". She is not the only one that noted this problem. According to Wiener: "We have deployed regulation for three decades with increasing *ex-ante* analysis of its predicted impacts, but too little *ex-post* analysis of its actual impacts".[49]

"Technologies of humility" can only be achieved by observing technological disasters from the past and the risk analysis and policy-relevant science so that the four focal points such as *framing*, *vulnerability*, *distribution*, and *learning* can be developed thus providing a "framework for the questions we should ask of almost every human enterprise that intends to alter society: what is the purpose; who will be hurt; who benefits; and how can we know?"[50]

To successfully regulate any social construct it is important to acknowledge every segment of it. Viskovic warned that not all social relations are subjected to legal paradigms, as often in modern society there is more to understand before regulating it: biological, economic, political, constitutional implications of a certain phenomenon. These implications Viskovic defines as "pre-normative legal relations" and as such, they become an important source of regulation before the concrete legal norms are created.[51] So to regulate we have to understand these pre-normative circumstances so that we can mobilize creativity so that knowing when, what, and how to regulate does not become "daunting", as Brownsword puts it, but rather a learning curve.

I believe that globalization has contributed to differing attitudes in understanding technology. Besides the remarkable similarities, but also differences that globalization brings, countries adopt a variance of observable legal models of regulation so some models of regulation may prioritize, for instance, religion over constitutional law or fundamental rights, or globalized free trade logic which relies on the trend of relaxation of state oversight. The market is about competition, consumption, growth of private profits, production and sacralization of private property, including intellectual property monopoly.

One of the objectives of my research is to explore how bioconstitutionalism, as defined by Jasanoff, and biotechnological regulatory designs are conditioned by neoliberal ideological underpinnings. I believe that those "pre-normative legal relations" are predominantly conditioned by the ideology that is behind the currents in the production of inequality. While Foucault understood neoliberalism

[46] S. JASANOFF, *Technologies of Humility: Citizen Participation in Governing Science*, cit. 238.
[47] *Ivi,* 239.
[48] *Ivi, 240*.
[49] J.B. WIENER, *The Regulation of Technology and the Technology of Regulation*, *Technology in Society* 26, 2004, 496.
[50] S. JASANOFF, *Technologies of Humility: Citizen Participation in Governing Science*, cit. 240.
[51] N. VISKOVIC, *Pojam Prava: Prilog Integralnoj Teoriji Prava*, Split, 1981, 83-94.

as an approach to human governance and a way of thinking and being,[52] for Harvey it is much more than that: "It has been part of the genius of neoliberal theory to provide a benevolent mask full of wonderful-sounding words like freedom, liberty, choice, and rights, to hide the grim realities of the restoration or reconstitution of naked class power, locally as well as transnationally, but most particularly in the main financial centres of global capitalism".[53] Rose observed that the projects to govern biotechnology is characterized be the alliances between political authorities and promise of capitalism as "the politics of the life sciences – the politics of life itself – has been shaped by those who controlled the human, technical and financial resources" to fund that scientific research.[54] The private corporations and foundations were involved in the capitalization of life science and thus "biopolitics becomes bioeconomics". In the section on legal implications of bioprinting technology I have mentioned the challenges, it poses to the concept of body ownership and commercialization of alienated body parts. Rose observed that new biotechnology brings novel molecular commodification. In his view the "classical distinction made in moral philosophy between that which is not human – ownable, tradeable, commodifiable – and that which is human – not legitimate material for such commodification – no longer seems so stable".[55] The health of citizens and their dependence on the biotechnological innovations is now conditioned by the very same principles that neoliberal free market thrives on: competition, the sanctity of intellectual property, competition, market discipline and circulation of commodities.

6. Conclusion

It should be noted, back in 1938 when the term neoliberalism has been coined the focus of liberal intellectual movement headed by Hayek was the redefinition of the functions of the state in market interventions opposing not only market socialism ideas developed by Oscar Lange but also Keynesian theory.[56] Cohen suggests that neoliberalism founders, in fact, did not indent to implement market ethics and principles of contract and consent on every feature of human life: "for it was the radicals of the 1960s that cleared away the very taboos surrounding the body that would have inhibited the newest possibilities of modern biotechnology".[57] And even so, this just suggests that neoliberal regulatory model did not resist biotechnological challenges as much of those advancements were a magnet for one of the creeds of the neoliberal markets: consumption. To move forward with the regulation of bioprinting we need to re-evaluate the relationship between technology and market (trade).

For Harari, we are living in an era of organisms shaped by intelligent design where *homo sapiens* is replaced by "superhumans".[58] And that is hardly a scene from the Westworld. Some scientists seem

[52] M. FOUCAULT, *The Birth of Biopolitics Lectures at the College de France*, New York, 2008, 218.
[53] D. HARVEY, *A Brief History of Neoliberalism*, epub, Oxford, 2005, 271.
[54] N. ROSE, *The Politics of Life Itself Biomedicine, Power and Subjectivity in the Twenty-First Century*, New Jersey, 2007, 31-34.
[55] *Ibidem.*
[56] D. HARVEY, *op.cit.* 59-61.
[57] E. COHEN, *Biotechnology and the Spirit of Capitalism*, in *New Atlantis,* 12, 2006, 62.
[58] Y.N. HARARI, *Sapiens: A Brief History of Humankind*, London, 2014, 8.

very well aware of this and suggest that "the only economic and reasonable way to commercialize organ-printing technology is to systematically employ scalable automated robotic technology and to build an integrated organ biofabrication line. It is not sufficient to develop just one robotic device – a bioprinter (it) will require the development of a series of integrated automated robotic devices or an organ biofabrication line".[59]

Should bioconstitutional relations between the state and our bodies be left at the mercy on neoliberal regulation creed we might as well accept that Ishiguro, Orwell and others prophesied our future.

The core values of documents such as Oviedo Convention must not be abandoned, and thus protection of our bodies via normative bioethics and principle of dignity, justice, informed consent and human rights might be uncomfortable notion for regulators who would leave bioprinting to the market. As the human body has an ontological value, we have to always remind decision-makers that the human body cannot and should be observed as a mere commodity. In doing so we should never forget that humans are embodied beings with natural limits, vulnerabilities and imperfections.

[59] V. MIRONOV et al., *Organ Printing: From Bioprinter to Organ Biofabrication Line*, in *Current Opinion in Biotechnology*, 22, 2011, 667.

ISSN 2284-4503

Special issue

Human germline genome editing and human rights law: A "brave new world" is not here to come

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ABSTRACT: Since the dawn of genetic engineering, potential application of genome editing on the germline, implying heritable DNA alterations, has been considered extremely controversial. Unsurprisingly, therefore, limits to human germline genome editing have been envisaged in international normative instruments in the'90s. Nowadays, the rise of new technical possibilities, like CRISPR-Cas9, urges for a due regulation of basic and pre-clinical research on gametes and embryos not destined to reproduction, while reinforcing the prohibition of clinical research and clinical applications on human beings. Keeping in mind the distinction between different kind of research, the paper addresses the contribution of human rights law in the debate on legitimacy of human germline genome editing.

KEYWORDS: Human germline genome editing; right to science; eugenics; UNESCO Declaration on the Human Genome and Human Rights; Oviedo Convention

1. Introduction

Limits and restrictions to human germline genome editing (HGGE) have been envisaged in international normative instruments in the late '90s, when the public debate was dominated by worries for potential drifts like those masterfully pictured by Huxley in 1932.[1] At the emergence of genetic engineering, predetermination of individuals' abilities and societal classification of people, according to different levels of intellectual enhancement, were feared as potential results of any intervention on the human genome.

Today – as CRISPR-Cas9 has translated into reality interventions that in past could be only pictured with a powerful imagination – different voices rise, calling for due regulation of basic and pre-clinical

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The article was peer-reviewed by the editorial committee.

[1] In *Brave new world*, written in 1931 and published in 1932, A. HUXLEY describes a World State, whose citizens are environmentally conditioned to fit into an intelligence-based social hierarchy. Although the novel does not refer to genetic manipulation, but rather to sleep-learning, psychological manipulation and Pavlonial or classical conditioning, it astonishingly anticipates scientific advancements in reproductive technology.

research, rather than a total ban.[2] According to many, precluding research on gametes and embryos not destined to implantation would impede to assess the scientific feasibility and the ethical acceptability of potential clinical applications of HGGE.[3] In fact, while it is currently too early to consider human genome editing for clinical reproductive purposes, some experts consider that – when all safety, efficacy and governance needs will be duly met – certain uses of HGGE in human reproduction might be considered morally acceptable and thus applied.[4]

The paper addresses the contribution of human rights law in the debate on legitimacy of human germline genome editing, clearly keeping in mind the distinction between, on the one hand, basic research[5] and pre-clinical research on gametes and embryos not destined to reproduction and, on the other, clinical research and clinical applications on human beings.

2. Human germline genome editing: total ban or rigorous restrictions?

Since the dawn of genetic engineering technologies, it became clear that potential application of genome editing on the germline might be extremely controversial. In fact, while somatic gene editing permits to target genes in selected cells of living patients and does not imply heritable DNA alterations, genome editing applied to human embryos or gametes involves the genetic modification of germ cells. This means that this technique affects all body cells of the future individuals, including their own gametes and that, as such, these DNA modifications will be inherited by their offspring (and by the offspring of their offspring).

Many Scholars consider that germline manipulation represents an ethical limit that should never be contravened, even when it will be eventually sufficiently safe.[6] Although the arguments supporting

[2] J. HALPERN et al., *Societal and Ethical Impacts of Germline Genome Editing: How Can We Secure Human Rights?*, in *The CRISPR Journal*, 2, 5, 2019, 293-298; G. DE WERT, G. PENNINGS, A. CLARKE et al., *Human germline gene editing. Recommendations of ESHG and ESHRE*, in *Human Reproduction Open*, 2018, 1-5.

[3] Italian Committee for Bioethics (ICB), *L'editing genetico e la tecnica CRISPR-CAS9: considerazioni etiche,* 2017, 5.1; available at: http://bioetica.governo.it/italiano/documenti/pareri-e-risposte/l-editing-genetico-e-la-tecnica-crispr-cas9-considerazioni-etiche/ (last visited 4/01/2021). While urging research on gene editing on human somatic cells, the Committee unanimously rejects experimentation on gametes and embryos intended to be used in reproduction. With regard to gene editing on gametes/embryos not destined to reproduction, the Committee could not reach a unanimous view: some members encourage basic and pre-clinical research, other consider that such research is not currently justified, because gamete selection is clinically preferable to gamete editing. They also stress that assessing the effectiveness and safety of *in vitro* gene editing on embryos is not possible, since results of the genetic modification can be assessed at birth or even later ; *ibidem*, 5.2.

[4] Hinxton Group Statement, *Statement on genome editing technologies and human germline genetic modification*, 2015, http://www.hinxtongroup.org/Hinxton2015_Statement.pdf (last visited 4/01/2021).

[5] Basic research can be defined as research done in laboratory, *in vitro*, aimed at improving scientific knowledge and "performed without thought of practical ends": V. BUSH, *Science: The Endless Frontier*, United States Government Printing Office, 1945, Chapter 3, available at https://www.nsf.gov/od/lpa/nsf50/vbush1945.htm#ch3.3 (last visited 4/01/2021).
See also N. ROLL-HANSEN, *Why the Distinction between Basic (Theoretical) and Applied (Practical) Research is Important in the Politics of Science*, London School of Economics, Centre for the Philosophy of Natural and Social Science Contingency and Dissent in Science Technical, 2009.

[6] K. DRABIAK, *The Nuffield Council's green light for genome editing human embryos defies fundamental human rights law*, in *Bioethics*, 34, 2020, 223-227; M. DARNOVSKY, L. LOWTHORP, K. HASSON, *Reproductive gene editing*

this view might vary, a major idea is that human genome deserve a special protection: to some extent it is "regarded as a 'natural' connection between all human beings, as it has been handed down to us by our predecessors".[7] This principle is traceable under Art. 1 of the Universal Declaration on the Human Genome and Human Rights, adopted by UNESCO in 1997, according which human genome represents "in a symbolic sense […] the heritage of humanity", as it "underlies the fundamental unity of all members of the human family, as well as the recognition of their inherent dignity and diversity".[8] In 1982, in a Recommendation on genetic engineering, the Parliamentary Assembly of the Council of Europe, addressing in general terms the concerns "arising from uncertainty as to the health, safety and environmental implications of experimental research", has deeply focused on the longer-term legal, social and ethical issues of the "prospect of knowing and interfering with a person's inheritable genetic pattern".[9] In this document, the plenary organ of the Council of Europe took a clear position against HGGE, stating that "the rights to life and to human dignity protected by Articles 2 and 3 of the European Convention on Human Rights imply the right to inherit a genetic pattern which has not been artificially changed".[10] Within the Council of Europe, the same approach has been confirmed later, with the adoption of the Convention for the protection of human rights and dignity of the human being with regard to the application of biology and medicine (Oviedo Convention).[11] Art. 13 of the Oviedo Convention states that "an intervention seeking to modify the human genome may only be undertaken for preventive, diagnostic or therapeutic purposes and only if its aim is not to introduce any modification in the genome of any descendants". According to someone, the reading of the norm could imply that "genome editing for therapeutic or preventive purposes where the modification in the genome of the descendants is not the aim but is incidental to the process, might still be in accordance with the Oviedo Convention";[12] nonetheless, the *ratio* of the provision seems to be indisputably focused on the ban of HGGE.

From that time, however, many scientific improvements had come, and the process is still ongoing.

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imperils universal human rights, in *OpenGlobalRights*, 15 February 2018, available at https://www.openglobalrights.org/reproductive-gene-editing-imperils-universal-human-rights/ (last visited 4/01/2021); E. ANDORNO, A.E. YAMIN, *The right to design babies? Human rights and bioethics,* in *OpenGlobalRights*, 8 January 2019, available at https://www.openglobalrights.org/the-right-to-design-babies-human-rights-and-bioethics/ (last visited 4/01/2021).

[7] N. PRIMC, *Do we have a right to an unmanipulated genome? The human genome as the common heritage of mankind*, in *Bioethics*, 34, 2020, 41-48, 41.

[8] The Universal Declaration on the Human Genome and Human Rights has been adopted at the 29th Session of the UNESCO General Conference on 11 November 1997. The United Nations General Assembly endorsed the Declaration by its Resolution 53/152 of 9 December 1998.

[9] Parliamentary Assembly of the Council of Europe (PACE), Recommendation 934 (1982) of 26 January 1982, *Genetic engineering*, para 2.

[10] *Ivi*, Art. 4 lett. a.

[11] Convention for the protection of human rights and dignity of the human being with regard to the application of biology and medicine (*Convention on Human Rights and Biomedicine*), Oviedo, 1997, European Treaty Series No.164.

[12] R. YOTOVA, *The Regulation of Genome Editing and Human Reproduction Under International Law, EU Law and Comparative Law*, Report to the Nuffield Council on Bioethics, June 2017, 4-5: available at https://www.nuffieldbioethics.org/wp-content/uploads/Report-regulation-GEHR-for-web.pdf (last visited 4/01/2021).

In 2020 the Nobel Prize in Chemistry has been awarded to Emmanuelle Charpentier and Jennifer Doudna for the development of CRISPR/Cas9. Commonly defined as "genetic scissors", this genome editing technology permits to change the DNA of animals, plants and microorganisms with extremely high precision and has therefore a revolutionary impact on life sciences, with potential applications to cancer or inherited diseases therapies. However, its application on human germline is considered highly controversial not only from a scientific perspective (as, in light of the current knowledge, it is still premature), but also from an ethical point of view. Unsurprisingly, therefore, when CRISPR-Cas9 was used to change the DNA of human embryos *in vitro* in 2015 for the first time, the disagreement about whether altering the genes of future generations should be permissible or not had strongly emerged.[13] More recently, the entire scientific community has been deeply shocked and unanimously expressed indignation and concern when, in November 2018, a Chinese biophysicist, He Jiankui, announced that two genetically modified twins had born. The embryos underwent a "genetic surgery", to become resistant to HIV infection, through the application of CRISPR/Cas9 technology, before being implanted in the maternal womb.

While the reaction of the scientific community was unanimous in blaming He Jiankui for having clearly ignored basic rules for research on humans, also violating norms and principles of medical practice, his irresponsive action also contributed to renovate the debate over the need to regulate the use of HGGE, rather than simply banning it.[14] In other words, as his misconduct proved that intervention on human germline is technically achievable and that unscrupulous and dishonest scientists may (easily) apply it, the question is whether, rather than simply placing a ban, basic and pre-clinical research should not be clearly regulated in order to make, in a near future, clinical research on humans safe, practicable and consistent with ethical standards.

If a large debate on possible regulation of human genome editing is not renounceable nor postponable nowadays, what role is played by human rights law?

3. Science and human rights law: supporting development and fixing boundaries

The effective safeguard of many fundamental human rights is strictly linked with progress in science and technology. It's undeniable, for example, that the enjoyment of the highest attainable standard of physical and mental health – protected, among other international provisions, under Art. 12 of the International Covenant on Economic, Social and Cultural Rights (ICESCR) – depends, in large part,

[13] National Academies of Sciences, *Engingeering and Medicine, Human Genome Editing: Science, Ethics, and Governance*, Washington DC, 2017; Nationale Akademie der Wissenschaften Leopoldina, Deutsche Forschungsgemeinschaft, acatech – Deutsche Akademie der Technikwissenschaften, Union der deutschen Akademien der Wissenschaften, *Chancen und Grenzen des genome editing/The opportunities and limits of genome editing*, 2015, available at: https://www.leopoldina.org/uploads/tx_leopublication/2015_3Akad_Stellungnahme_Genome_Editing_01.pdf (last visited 4/01/2021); Koninklijke Nederlandse Akademie van Wetenschappen, *Genome Editing. Position Paper of the Royal Netherlands Academy of Arts and Sciences*, 2016, available at: https://knaw.nl/en/news/publications/genome-editing (last visited 4/01/2021).

[14] G. Q. Daley, R. Lovell-Badge, J. Steffann, among others, consider that halting responsible research would be unwise: *After the Storm: A Responsible Path for Genome Editing*, in *New Eng. J. Med.*, 380, 2019, 897-899.

from scientific development.[15] Moreover, science not only offers solutions to individual, social, economic, and developmental issues, but it has also an autonomous standing among other fundamental rights: Art. 15 ICESCR enshrines the right "to enjoy the benefits of scientific progress and its applications", as well as the "the freedom indispensable for scientific research and creative activity".[16] Nonetheless, while scientific development has a pivotal role in supporting the ability of people to "conceive of a better future that is not only desirable but attainable",[17] at the same time, human rights law sets boundaries that science and technology can't cross and provides guidance to balance conflicting interests. Indeed, human rights identify limits bearing upon States in regulating research on human germline genome editing.

3.1 Regulating HGGE to realize the right to science

The fundamental right to science represents a multifaceted normative basis supporting research on HGGE. Different components of this fundamental right are relevant: not only the right to enjoy the benefits of scientific progress *per se*, but also the freedom of research, as well as the right to participate in science. The three dimensions, in particular, fit perfectly with basic and pre-clinical research on human germline genome editing.

As clarified in the Venice Statement, benefits of scientific progress "encompass not only scientific results and outcomes but also the scientific process, its methodologies and tools".[18] On the same line, the General Comment n. 25 on the right to science – adopted in 2020 by the Committee on economic social and cultural rights – states that science benefits include "the scientific knowledge and information directly deriving from scientific activity".[19] The right to enjoy the benefits of scientific progress, therefore, certainly covers the general enhancement of the conditions for further scientific activity, realized through basic and pre-clinical research on HGGE.

Freedom of research is the second major component of the right to science: according to General Comment n. 25, "in order to flourish and develop, science requires the robust protection of freedom of research".[20] As clearly stated in Art. 15 para 3 ICESCR, this freedom is "indispensable", although

[15] International Covenant on Economic, Social and Cultural Rights adopted by General Assembly resolution 2200A (XXI) of 16 December 1966.

[16] A similar provision is contained under Art. 27 of the *Universal Declaration of Human Rights*, 1948.

[17] Human Rights Council, *Report of the Special Rapporteur in the Field of Cultural Rights, Farida Shaheed on the Right to Enjoy the Benefits of Scientific Progress and its Applications*, 14 May 2012 UN Doc. A/ HRC/20/26, para. 20.

[18] Venice Statement on the Right to Enjoy the Benefits of Scientific Progress and its Applications, para 8. Such Statement is the outcome of three expert meetings held between June 2007 and July 2009 under the auspices by UNESCO in collaboration with the Amsterdam Centre for International Law, the Irish Centre for Human Rights, and the European Inter-University Centre for Human Rights and Democratization.

[19] UN Committee on economic social and cultural rights, *General comment No. 25 (2020) on science and economic, social and cultural rights (article 15 (1) (b), (2), (3) and (4) of the International Covenant on Economic, Social and Cultural Rights*. 30 April 2020, UN Doc. E/C.12/GC/25, para 8, available at: https://documents-dds-ny.un.org/doc/UNDOC/GEN/G20/108/12/PDF/G2010812.pdf?OpenElement (last visited 4/01/2021).

[20] *Ivi*, para 13. According to the UN Committee on economic social and cultural rights, "this freedom includes, at the least, the following dimensions: protection of researchers from undue influence on their independent judgment; the possibility for researchers to set up autonomous research institutions and to define the aims and objectives of the research and the methods to be adopted; the freedom of researchers to freely and openly

not unlimited. Limits can be set, in particular, as long as science and its applications interfere with other economic, social and cultural rights, but most importantly, boundaries are to be fixed whenever "the research affects human beings", with the aim "to protect their dignity, their integrity and their consent when involved in the research".[21] The need to support freedom of research in the field of human genome emerges also form the provision of Art. 14 of the UNESCO Declaration on the Human Genome and Human Rights, which states that "States should take appropriate measures to foster the intellectual and material conditions favorable to freedom in the conduct of research on the human genome and to consider the ethical, legal, social and economic implications of such research […]".

Last but not least, the *enjoyment* of the benefits of scientific progress covers not only the dissemination of its outcomes, but also the participation in its development. Along with the freedom of research at the benefit of scientists, the right to science includes also the right of anyone to take part in science, that is – in the words of the Special Rapporteur in the field of cultural rights – the "participation of individuals and communities in decision-making" and, more in general, "opportunities for all to contribute to the scientific enterprise".[22] General Comment n. 25 clearly identifies this right to take part in scientific progress and in decisions concerning its direction, clarifying that the benefits of scientific advancement include "the development of the critical mind and faculties associated with doing science".[23] Therefore, if it is clear that "the incorporation of any kind of technological innovation, including genetic therapies, requires broad collective deliberation regarding its permissible uses, foreseeable effects, and regulation",[24] it is also true that the improvement of knowledge (through basic and pre-clinical research) might provide the necessary instruments and tools to involve people in a fruitful dialogue on potential clinical applications, ethical limits and normative standards for HGGE.

Indeed, the three components of the right to science militate in favor of a (proper) regulation of basic and pre-clinical research on HGGE, while a total ban (but also practical impediments like the prohibition to use supernumerary embryos in research)[25] could imply a violation of Article 15 of the ICESCR, interfering with scientific progress and impairing the right of many people to enjoy the benefits of science and its applications.

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question the ethical value of certain projects and the right to withdraw from those projects if their conscience so dictates; the freedom of researchers to cooperate with other researchers, both nationally and internationally; and the sharing of scientific data and analysis with policymakers, and with the public wherever possible".

[21] *Ivi*, para 22.

[22] Human Rights Council, *Report of the Special Rapporteur in the Field of Cultural Rights cit*., para. 25. See also UN Committee on economic social and cultural rights, *General comment No. 25* cit., para 9-11.

[23] UN Committee on economic social and cultural rights, *General comment No. 25* cit., para 10.

[24] E. ANDORNO, A.E. YAMIN, *op. cit*.

[25] L. POLI, *The Regulation of Human Germline Genome Modification in Italy*, in A. BOGGIO, C. ROMANO, J. ALMQVIST (Eds.), *Human Germline Genome Modification and the Right to Science: A Comparative Study of National Laws and Policies*, Cambridge, 2020, 335-357.

3.2 Setting limits in the name of human dignity

Current human rights law does not contain a complete ban on genome editing; it rather envisages limits that States must observe in regulating scientific research in this field.

Most importantly, Art. 13 of the Oviedo Convention, as already mentioned, prohibits any clinical research and application of human germline genome editing. Along with other provisions contained in Chapter IV of the Convention, the norm is clearly inspired by the precautionary principle and, as explained in the Explanatory Report of the Oviedo Convention, it aims at preventing misuses of scientific progress in the field of genome editing that "may endanger not only the individual but the species itself", through the "intentional modification of the human genome so as to produce individuals or entire groups endowed with particular characteristics and required qualities".[26]

Another clear ban concerns the reproductive cloning of human beings. This practice is outlawed by Art. 1 of in the Additional Protocol to the Oviedo Convention on the prohibition of cloning human beings,[27] by Art. 11 of the UNESCO Declaration on human genome as well as by Art. 3 of the Charter of fundamental rights of the European Union.[28] The Explanatory Report of the Additional Protocol to the Oviedo Convention clarifies that "deliberately cloning humans is a threat to human identity, as it would give up the indispensable protection against the predetermination of the human genetic constitution by a third party".[29] Such prohibition is clearly based on the need to preserve human dignity, which is considered to be "endangered by instrumentalization through artificial human cloning".[30] Indeed, "it is in the interest of all persons to keep the essentially random nature of the composition of their own genes", because "naturally occurring genetic recombination is likely to create more freedom for the human being than a predetermined genetic make-up".[31]

The prohibitions of clinical research and application of HGGE and of reproductive cloning of human beings are grounded on similar fears: the human species as well as humanity might be put in danger through the application of techniques aimed at predefining genetic characteristics of individuals. In other words, these bans aim at safeguarding a "collective dimension" of human dignity, namely the "long-term interests of society, future generations, and humankind" that go beyond individual rights "of prospective parents and […] future offspring".[32]

Two major concerns regard eugenics and enhancement.

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[26] Explanatory Report to the Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, para. 89.

[27] Additional Protocol to the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine, on the Prohibition of Cloning Human Beings, Paris, 1998, European Treaty Series No. 168.

[28] *Charter of fundamental rights of the European Union* (2000/C 364/01), Nice, 2000.

[29] Explanatory Report to the Additional Protocol to the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine, on the Prohibition of Cloning Human Beings, para 3.

[30] *Ibidem*.

[31] *Ibidem*.

[32] B.C. VAN BEERS, *Rewriting the human genome, rewriting human rights law? Human rights, human dignity, and human germline modification in the CRISPR era*, in *Journal of Law and the Biosciences*, 2020, 35-36.

Eugenics as a practice "aiming at the selection of persons", is explicitly prohibited under Article 3.2 of the Charter of Fundamental Rights of the European Union, which affirms the right to the integrity of the person in the fields of medicine and biology. According to the Explanations Relating to the Charter of Fundamental Rights, Article 3.2 refers to "possible situations in which selection programmes are organized and implemented, involving campaigns for sterilization, forced pregnancy, compulsory ethnic marriage among others, all acts deemed to be international crimes in the Statute of the International Criminal Court adopted in Rome on 17 July 1998".[33] While the intention of the drafters of the EU Charter was to ban projects aimed at the improvement of the human race, based on the selection by the State of who can procreate, it is true that HGGE might allow a new form of eugenics. Intended parents can know in advance the genetic makeup of their future children, and through genetic manipulation, they could make decisions about their offspring's genes in view of an improvement that goes beyond the treatment of medical disorders.

Enhancement, then, represents the second major concern about misapplication of HGGE. The potential use of germline editing to enhance traits, rather than to serve therapeutic needs, poses serious moral issues and risks to exacerbate social inequities. Additionally, it is true that setting the dividing line between healing and enhancement might not be easy in practice.[34]

As far as basic and pre-clinical research are concerned, some rules delineate general limits, mainly identifying the respect for human rights, fundamental freedoms and human dignity of individuals as prevailing over science *per se* (Art. 10 UNESCO Declaration on human genome, Art. 2 Oviedo Convention), or imposing rigorous and prior assessment of the potential risks and benefits for any research, treatment or diagnosis affecting an individual's genome (Art. 5 lett. a UNESCO Declaration on human genome). In addition, many provisions stress the key importance of free and informed consent of the person(s) involved in the research (Art. 3 Charter of fundamental rights of the EU, Art. 5 Oviedo Convention, Art. 5 UNESCO Declaration on human genome). Even with regard to basic and pre-clinical research, informed consent of people whose gametes or supernumerary embryos are to be used in research remain of crucial importance.

A stricter limit to basic and pre-clinical research is stated under Art. 18 of the Oviedo Convention, according which "[t]he creation of human embryos for research purposes is prohibited". The provision also recommends that national law ensure adequate protection of the embryos when *in vitro* research on them is allowed. Hence, to the extent that it is not prohibited, basic research on (supernumerary) embryos is to be considered permitted pursuant to the Oviedo Convention, despite the extreme cautious approach adopted in the Explanatory Report, where it is stressed that Art. 18 "does not take a stand on the admissibility of the principle of research on *in vitro* embryos".[35]

A final set of rules which appears relevant in this field of research are those concerning the respect for genetic diversity and/or the prohibition of genetic discrimination.[36] In particular, Art. 2 of the UNESCO Declaration on human genome provides that "[e]veryone has a right to respect for their

[33] Explanations Relating to the Charter of Fundamental Rights, 2007, OJ C 303.

[34] See B.C. VAN BEERS, *op. cit.*, 20-24.

[35] Expl rep 116

[36] See I.V. MOTOC, *The International Law of Genetic Discrimination: The Power of "Never Again",* in T. MURPHY (Ed.), *New Technologies and Human Rights*, Oxford, 2009, Collected Courses of the Academy of European Law, XVII/2, 222-245.

ISSN 2284-4503

dignity and for their rights regardless of their genetic characteristics. That dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity". In addition, according to Art. 6 of the same document, "[n]o one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity". Similarly, the Oviedo Convention bans all forms of discrimination based on a person's genetic make-up (Art. 11), allowing predictive genetic tests only for health or scientific research purposes (Art. 12). Additional documents have been adopted by the UN Economic and Social Committee[37] and the World Health Organization.[38] Finally, the 2003 UNESCO International Declaration on Human Genetic Data details principles that should govern the collection, processing, use and storage of human genetic data.[39] In particular, Art. 7 (entitled "[n]on-discrimination and non-stigmatization") states that "[e]very effort should be made to ensure that human genetic data and human proteomic data are not used for purposes that discriminate in a way that is intended to infringe, or has the effect of infringing human rights, fundamental freedoms or human dignity of an individual or for purposes that lead to the stigmatization of an individual, a family, a group or communities". With regard to HGGE, these norms will be probably relevant only when (and if) this technique will be concretely used in clinical research and practice. Moreover, the protection against genetic discrimination would be probably better applicable to individuals who do not benefit from access to HGGE, rather than to those who can use this technology to avoid genetic diseases. In any case, principles concerning genetic diversity and the prohibition of genetic discrimination still define some boundaries to basic and pre-clinical research, to the extent they consider genetic variety as an element of richness for humanity deserving special protection, net of therapeutic intervention on human genome to correct serious hereditary diseases.

4. Concluding remarks: defining values for "good" science

Pervasive individual and public interest in genetics will probably not allow unlimited impediments to the improvement of knowledge on human germline genome editing. In fact, any arbitrary obstacle to scientific developments in this field would certainly result in a violation of the right to science. While scientists' freedom of research is not without bounds, we all would significantly benefit from a deeper understanding of genome editing, also to be able to actively participate in the decision-making process, defining standards and limits for HGGE.

The Parliamentary Assembly of the Council of Europe has recognized in 2017 that, while the Oviedo Convention prohibits intervention on germline genome, nothing precludes a possible amendment of

[37] ECOSOC, Resolution 2001/39 on Genetic Privacy and Non-Discrimination, adopted on 26 July 2001. The ECOSOC urges States to ensure that no-one shall be subjected to discrimination based on genetic characteristics and to take measures to prevent the use of genetic information and testing leading to discrimination or exclusion against individuals, particularly in social, medical or employment-related areas, whether in the public or the private sector.

[38] Council for International Organizations of Medical Sciences and World Health Organization, *International Ethical Guidelines for Biomedical Research Involving Human Subjects*, 2016.

[39] The International Declaration on Human Genetic Data was adopted at UNESCO's 32nd General Conference on 16 October 2003.

the treaty.[40] While urging "member States which have not yet ratified the Oviedo Convention to do so without further delay, or, as a minimum, to put in place a national ban on establishing a pregnancy with germline cells or human embryos having undergone intentional genome editing",[41] it has also explicitly encouraged the development of a common regulatory and legal framework to balance "potential benefits and risks of these technologies aiming to treat serious diseases, while preventing abuse or adverse effects of genetic technology on human beings".[42]

It is true that, along with potential benefits of intervening on gametes or on early-stage embryos to treat genetic diseases, germline modifications might leave room for abuses; however, concerns about eugenics and enhancement do not refer to the technique *per se*, but rather to its possible misuses. In this perspective, HGGE is not different from other scientific inventions: science in not inherently good. Along with technology, it is rather a vehicle serving whatever values and ideals it is guided by. Indeed, the law "will be the custodian of our values as we decide on the right uses of genetic technologies and knowledge".[43] The discussion on the legitimacy of human germline genome editing needs to be contextualized within the human rights framework, which will certainly contribute to trace the dividing line between Nobel prize winners and irresponsible (potentially criminal) scientists.

[40] Parliamentary Assembly of the Council of Europe (PACE), Recommendation 2115 (2017) of 12 October 2017, *The use of new genetic technologies in human beings*, para 3.
[41] *Ivi*, para 5.1.
[42] *Ivi*, para 5.2.
[43] I.V. MOTOC, *op. cit.*, 225.

Decisioni in materia di *editing* genetico e partecipazione democratica. Un ambito privilegiato per l'applicazione dei principi della democrazia deliberativa?

Giada Ragone *

GENE EDITING DECISIONS AND DEMOCRATIC PARTICIPATION: A PRIVILEGED AREA FOR THE APPLICATION OF THE PRINCIPLES OF DELIBERATIVE DEMOCRACY?

ABSTRACT: The frontiers reached by gene editing and their applications for animals, plants and humans raise numerous tricky issues, posing new and renewed challenges for constitutional law scholars. This paper focuses on the role that the public can play in decision-making processes aimed at regulating gene-editing technologies. Thus, it reflects on the opportunity to develop and enhance deliberative arenas, which guarantee a fuller participation of citizens in legislative choices that touch upon controversial scientific issues of general interest, in compliance with the principles of deliberative democracy.

KEYWORDS: deliberative democracy; participatory democracy; deliberative arenas; gene editing; public participation

SOMMARIO: 1. Introduzione – 2. Deliberativismo e democrazia partecipativa: un inquadramento teorico e i modelli a cui guardare – 3. La manipolazione genetica come ambito privilegiato per la partecipazione pubblica – 4. Il caso della *Global Citizens' Assembly on Genome Editing*. – 5. Conclusioni.

1. Introduzione

Nel corso del 2020, il *Centre for Deliberative Democracy and Global Governance* dell'Università di Canberra (Australia) ed una rete internazionale di ricercatori hanno annunciato la convocazione della prima "assemblea cittadina globale" in materia di *genome editing*[1]. L'iniziativa consiste nella realizzazione di un *forum* in cui cittadini provenienti da tutto il mondo possano discutere, scambiarsi opinioni e ricevere informazioni circa gli sviluppi e le implica-

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[1] Cfr. N. CURATO, S. NIEMEYER, *Why we need a global citizens' assembly on gene editing*, in *TheConversation.com* , 17 settembre 2020; J.S. DRYZEK et al., *Global citizen deliberation on genome editing, in American Association for the Advancement of Science*, 369, 6510, 18 settembre 2020, 1435-1437; J.S. DRYZEK, A. BÄCHTIGER, K. MILEWICZ, *Toward a Deliberative Global Citizens' Assembly,* in *Global Policy*, 1, 2011, 33-42; J. CHADWICK, *Plumbers and teachers are invited to share their views on designer babies and genetically enhanced potatoes as part of a citizens' assembly on 're-engineering' the human species*, in *Daily Mail Online*, 18 settembre 2020. Il sito ufficiale del progetto "Global Citizens' Assembly on Genome Editing. Connecting citizens, science and global governance" è: https://www.globalca.org/.

zioni delle più avanzate tecniche di ingegneria genetica, le quali consentono oggi di alterare la sequenza genetica di qualsiasi organismo (vegetale, animale e umano) «to produce favourable characteristics or remove unwanted ones»[2]. Al termine della fase di discussione, è chiesto all'assemblea di produrre delle raccomandazioni («moral and political regulation»[3]) che possano essere prese in considerazione dai soggetti che, a tutti i livelli, sono deputati alla regolamentazione delle suddette tecnologie.

Per decenni il dibattito giuridico sulla manipolazione genetica ha avuto ad oggetto principalmente la produzione di cibi OGM, ossia prodotti agricoli destinati all'alimentazione umana e animale, il cui materiale genetico viene modificato in modo diverso da quanto si verifica in natura. Tuttavia le tecniche di alterazione del DNA che negli anni sono state sviluppate non riguardano la sola modificazione genetica di semi e varietà vegetali, ma coinvolgono tutte le forme di vita presenti sul pianeta, uomo incluso[4].

Una delle tecnologie più promettenti e versatili in questo ambito è quella nota come sistema CRISPR-Cas[5], la quale consente di tagliare e sostituire sequenze genomiche in maniera relativamente semplice, applicando un sistema che è naturalmente presente nei batteri e in altri organismi unicellulari. La "correzione" genetica può intervenire sulla linea germinale di un organismo, ossia prima che avvenga in esso la differenziazione cellulare, oppure più avanti nel suo sviluppo, sulla linea somatica. Laddove applicata alle cellule germinali, la modificazione non produce effetti sul solo organismo su cui si effettua ma verrà trasmessa a tutti gli organismi che in seguito riceveranno il suo patrimonio genetico. Pertanto, la manipolazione sulla linea germinale comporta la possibilità che effetti collaterali e imprevedibili dell'alterazione si trasformino in mutazioni genetiche permanenti[6].

Pur a fronte di queste ombre, è indubbio che le potenzialità del CRISPR siano vastissime e promettenti. Esse muovono dal miglioramento dei cibi ad applicazioni in ambito medico di enorme portata (si parla della possibilità di interrompere la diffusione di malattie come la malaria intervenendo sul DNA degli insetti che ne sono portatori, di aiutare la lotta al cancro, di curare rare malattie genetiche etc.), sino all'alterazione di embrioni umani prodotti *in vitro* per rimuovere dal patrimonio genetico dei nascituri eventuali geni considerati determinanti nello sviluppo di malattie o caratteristiche indesiderate. Risale, ormai, al 2018 la notizia che il CRISPR sarebbe stato impiegato per dar vita ai primi due essere umani geneticamente modificati nel controverso e opaco esperimento del ricercatore cinese Jiankui He: secondo quanto dichiarato dal medesimo scienziato, il *trial* clinico da lui condotto

[2] N. CURATO, S. NIEMEYER, *op. cit*., 1.

[3] J.S. DRYZEK et al., *Global citizen deliberation on genome editing*, cit., 1435.

[4] Al 2015 risale un primo celebre tentativo di modificazione genetica di embrioni umani, cui è seguita la convocazione di un summit internazionale di studiosi (l'International Summit on Human Gene Editing) a Washintong, concepito come «an initial attempt to keep the discussion about human genome editing thematically broad and open to input from a variety of stakeholders» (A. BLASIMME*, Why Include the Public in Genome Editing Governance Deliberation?*, in *AMA Journal of Ethics*, 21, 12, 2019, 1067).

[5] Acronimo di Clustered Regularly Interspaced Short Palindromic Repeats-Cas. Sulla tecnica in questione cfr. https://bit.ly/3gBT8b7 .

[6] Per questo, come ricordato in S. BONOMELLI, *Gene editing embrionale: il vaso di pandora è stato scoperchiato? Riflessioni a margine del caso di Jiankui He*, in *BioLaw Journal*, 3, 2019, 72, sia la *Convenzione di Oviedo* sia la *Dichiarazione universale sul genoma umano e i diritti umani UNESCO* vietano in modo assoluto gli interventi genetici sulla linea germinale.

avrebbe coinvolto coppie di aspiranti genitori composte da padri positivi all'HIV e avrebbe portato alla produzione di embrioni resistenti al virus in questione tramite la disattivazione del gene che solitamente permette il contagio negli essere umani[7]. L'operazione di *editing* genetico sarebbe stata, dunque, finalizzata ad evitare che i bambini nati dall'esperimento possano in futuro contrarre a propria volta l'HIV[8].

È del tutto evidente che le frontiere cui il *gene editing* è pervenuto e gli orizzonti verso cui simili tecnologie muovono sollevano numerose e delicate questioni, oltre che sul piano etico e scientifico, su quello giuridico, ponendo nuove e rinnovate sfide agli studiosi del diritto costituzionale[9]. Tra le molte, nel presente scritto si è scelto di riflettere, in particolare, sul ruolo che il pubblico può giocare nei processi decisionali volti alla regolamentazione delle descritte tecnologie. Si intende, così, offrire un contributo al dibattito circa l'opportunità di sviluppare e valorizzare arene deliberative[10] che, come quella convocata dagli studiosi dell'Università di Canberra, garantiscano una più piena partecipazione del pubblico a scelte normative che toccano questioni scientifiche controverse[11] di generale interesse, in ossequio ai principi della c.d. democrazia deliberativa[12].

A tal fine si procederà, preliminarmente, ad un inquadramento del deliberativismo e si cercherà di individuare i modelli di democrazia partecipativa che appaiono maggiormente adeguati ad essere presi in considerazione per decisioni *science and technology based*. Si chiarisce, peraltro, sin d'ora che «i modelli adottati sono assai vari, per cui non a una modellistica standardizzata è possibile riferirsi ma ad alcuni principi orientativi»[13].

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[7] Sull'esperimento in questione, cfr. S. BONOMELLI, *op. cit.,* 67 ss.

[8] L'esperimento sembrerebbe, tuttavia, in larga parte fallito. Cfr. D. PERRIN, G. BURGIO, *China's failed gene-edited baby experiment proves we're not ready for human embryo modification*, in *TheConversation.com*, 9 dicembre 2019.

[9] Sulle «interazioni tra i due "codici" (genetica e diritto) che compongono il "palinsesto della vita" nel processo di riconfigurazione "biocostituzionale" dei diritti» (M. TALLACCHINI, *Scienza e diritto. Prospettive di co-produzione*, in *Rivista di filosofia del diritto*, 2, 2012, 317) si veda, ampiamente, S. JASANOFF (a cura di), *Reframing Rights. Bioconstitutionalism in the Genetic Age*, Cambridge, 2011.

[10] Sul concetto, notoriamente L. BOBBIO, *Le arene deliberative*, in *Rivista Italiana di Politiche Pubbliche*, 3, 2002, 5 ss. e più recentemente V. MOLASCHI, *Le arene deliberative. Contributo allo studio delle nuove forme di partecipazione nei processi di decisione pubblica*, Napoli, 2018 e ID., *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico*, in *Nuove Autonomie*, 3, 2017, 464. In quest'ultima sede delle arene deliberative si parla come di «strumenti, ispirati ai principi della democrazia deliberativa, che sempre più spesso vengono utilizzati nel caso di decisioni pubbliche che riguardano questioni scientificamente e tecnicamente complesse, tipicamente nei settori della salute e della ricerca biomedica, delle biotecnologie e dell'ambiente».

[11] Per la definizione di "questione scientifica controversa" il rimando è d'obbligo a L. VIOLINI, *Le questioni scientifiche controverse nel procedimento amministrativo*, 1986, Pavia.

[12] A riguardo, nella letteratura italiana, cfr. inter alia, U. ALLEGRETTI, *Basi giuridiche della democrazia partecipativa in Italia: alcuni orientamenti*, in *Democrazia e diritto*, 3, 2006, 151 ss. e ID., *Democrazia partecipativa*, in *Enciclopedia del Diritto – Annali IV*, Milano, 2011, 295 ss.; R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa. Relazione al Convegno "La democrazia partecipativa in Italia e in Europa: esperienze e prospettive"* - Firenze, 2-3 aprile 2009, in *Rivista Astrid* e ID., *Democrazia deliberativa e principio di realtà*, in *Federalismi.it*, numero speciale 1, 2017, 1 ss.; A. FLORIDIA, *La democrazia deliberativa. Teorie, processi e sistemi*, Carocci, Roma, 2013 e ID., *Democrazia partecipativa e democrazia deliberativa: una risposta plausibile alla "crisi della democrazia"?,* in www.fondazionefeltrinelli.it, 29 marzo 2019.

[13] U. ALLEGRETTI*, Basi giuridiche della democrazia partecipativa in Italia: alcuni orientamenti,* cit.*,* 152.

In secondo luogo, si realizzerà una disamina delle caratteristiche che fanno dell'ambito prescelto, ossia l'*editing* genetico, un banco di prova particolarmente interessante per l'applicazione delle teorie deliberativiste.
Da ultimo, prima di volgere alle conclusioni, si sottolineeranno pregi e limiti della *Global Citizens' Assembly on Gene Editing*, ponendo particolare attenzione sulla scelta di fare un esperimento di dimensione globale.

2. Deliberativismo e democrazia partecipativa: un inquadramento teorico e i modelli a cui guardare

Solitamente, nella descrizione degli istituti e dei meccanismi che connotano le nostre democrazie contemporanee si fa ricorso ai concetti di "democrazia rappresentativa" e di "democrazia diretta". Se si prende ad esempio la Costituzione italiana, in essa si trova delineato uno Stato democratico che affida le principali funzioni decisionali ad organi statali che *rappresentano* il popolo e ad esso sono collegati tramite meccanismi elettivi diretti o indiretti; al contempo, sono presenti altresì istituti di c.d. democrazia diretta, come i *referendum*, l'iniziativa legislativa popolare e la petizione, che consentono al corpo elettorale di prendere decisioni che incidono sull'ordinamento *senza mediazione* di rappresentanti.
La realizzazione di istituti di democrazia diretta all'interno di sistemi rappresentativi risponde all'esigenza di «assicurare la partecipazione popolare alle decisioni che riguardano l'intera collettività e di colmare la distanza tra il popolo e l'apparato statuale»[14]. Ad un obiettivo non dissimile, seppure attraverso modalità differenti[15], tendono quegli istituti o procedure che la dottrina inquadra come esempi di "democrazia partecipativa" e che affondano le proprie radici nelle diverse teorie di "democrazia deliberativa"[16]. Non a caso la diffusione di strumenti tipici di queste accezioni di democrazia è considerata, *inter alia*, un portato della crisi della democrazia rappresentativa[17].
Prima di procedere ad un inquadramento definitorio, sono necessari due chiarimenti. Anzitutto, si concorda con l'opinione secondo cui «Democrazia Partecipativa e Democrazia Deliberativa non sono forme *alternative* alla democrazia rappresentativa; piuttosto sono modelli o idee che possono indicare alcune caratteristiche e segnare la maggiore o minore *qualità* della stessa democrazia rappresentativa»[18]. Gli istituti e le forme che vengono generalmente ricondotti nell'alveo di questi concetti

[14] R. BIN, G. PITRUZZELLA, *Diritto costituzionale*, XXI ed., Torino, 2020, 75.
[15] Così L. BOBBIO, G. POMATTO, *Il coinvolgimento dei cittadini nelle scelte pubbliche*, in *Meridiana: rivista di storia e scienze sociali*, 58, 2007, 46. Contra v. G. SCOFFONY, *La démocratie participative dans les États fédérés américains,* in F. ROBBE (a cura di), *La démocratie participative*, Parigi, 2007, 98, secondo cui sono riconducibili a forme di democrazia partecipativa anche gli istituti di democrazia diretta.
[16] Sulla possibilità che l'implementazione di forme di democrazia partecipativa offra una risposta credibile al bisogno di partecipazione dei cittadini nella vita della società e delle istituzioni, cfr. U. ALLEGRETTI, *Basi giuridiche della democrazia partecipativa in Italia: alcuni orientamenti,* cit., 151.
[17] Sul punto cfr. V. MOLASCHI, *Le arene deliberative, cit.,* 18-19; G. PEPE, *Il modello della democrazia partecipativa tra aspetti teorici e profili applicativi un'analisi comparata*, Padova, 2020, 13 ss. e A. FLORIDIA, *Democrazia partecipativa e democrazia deliberativa: una risposta plausibile alla "crisi della democrazia"?,* cit., passim.
[18] A. FLORIDIA, *Democrazia partecipativa e democrazia deliberativa: una risposta plausibile alla "crisi della democrazia"?,* cit., 2.

possono, dunque, essere concepiti come complementari rispetto alle istituzioni di democrazia rappresentativa e a quelle di democrazia diretta[19].

Il secondo chiarimento concerne il rapporto tra democrazia deliberativa e partecipativa: sebbene esistano ricostruzioni divergenti, in questa sede si aderisce all'impostazione secondo cui le dottrine deliberativiste[20] costituiscono la base teorica in cui affondano le proprie fondamenta le prassi riconducibili alla democrazia partecipativa[21].

Offrire una definizione univoca del deliberativismo non è cosa semplice: esso, infatti, costituisce una base, o «sfondo teorico»[22], molto variegato. Tuttavia è possibile rintracciare due caratteristiche che fungono da minimo comun denominatore alle sue varie teorizzazioni[23]: *in primis*, si tratta di un concetto di democrazia in cui i processi di decisione pubblica debbono prevedere una fase di scambio di opinioni e informazioni il più completo e imparziale possibile; *in secundis*, la discussione (o *deliberation*) deve essere inclusiva, coinvolgendo idealmente tutti i soggetti interessati dalla decisione finale o almeno un campione rappresentativo.

Quanto alle forme che può assumere la democrazia partecipativa, molte sono le prassi che sono state sperimentate in tutto il mondo, sia a livello nazionale sia a livello locale[24]: basti pensare che in letteratura sono state contate più di cento diverse tipologie di meccanismi partecipativi[25]. Tra i modelli più noti e diffusi vi sono le *citizens' juries*, i *deliberative polls*, i *town meetings*, i *Planungszellen*, i bilanci partecipati e le *consensus conference*[26]. Queste ultime, in particolare, rilevano per il tema del presente contributo, dal momento che costituiscono «un tipico esempio di processo partecipativo volto alla democratizzazione delle decisioni pubbliche c.d. *science and technology based*»[27]. Esse, infatti, mettono in dialogo esperti di una determinata questione tecnico-scientifica con un campione

[19] Cfr. U. ALLEGRETTI*, Basi giuridiche della democrazia partecipativa in Italia: alcuni orientamenti,* cit., 153; M. SETÄLÄ, *Connecting deliberative mini-publics to representative decision making*, in *European Journal of Political Research*, 56, 2017, 846 ss. Un interessante – ancorché non privo di criticità – connubio tra strumenti di democrazia diretta e di democrazia partecipativa è stato realizzato, in più occasioni, in Irlanda, dove importanti referendum su temi eticamente sensibili sono stati anticipati da assemblee deliberative: cfr. E. CAROLAN, *Ireland's Citizens' Assembly on Abortion as a Model for Democratic Change? Reflections on Hope, Hype and the Practical Challenges of Sortition*, in *IACL-AIDC Blog*, 28 novembre 2018; C. O' CINNEIDE, *The Citizens' Assembly Viewed in External Perspective: Useful, but not a Deliberative Deus Ex Machina*, in *IACL-AIDC Blog*, 12 dicembre 2018.

[20] La prima teorizzazione del concetto di democrazia deliberativa si trova in J.M. BESSETTE, *Deliberative Democracy: The Majority Principle in Republican Government*, in R. GOLDWIN, W.A. SCHAMBRA (a cura di), *How Democratic is the Constitution?*, Washington, 1980, 102 ss.

[21] Così R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa*, cit*.,* 2 ss.

[22] *Idem*, 3.

[23] Cfr. V. MOLASCHI, *Le arene deliberative,* cit*.,* 34 ss. Secondo Pellizzoni, che parla della democrazia deliberativa come della «innovazione più significativa nel dibattito e nelle pratiche democratiche degli ultimi decenni» (L. PELLIZZONI, *Comunità, partecipazione e democrazia deliberativa: un'esperienza italiana*, in *Quaderni di sociologia*, 68, 2015, 149), l'opzione chiave dell'ideale deliberativo consiste nell'instaurazione di una discussione organizzata.

[24] Per un interessante esempio di arena deliberativa organizzata a livello comunale cfr. G. BALDUZZI, D. SERVETTI (a cura di), *Discutere e agire. Una sperimentazione di democrazia deliberativa a Novara*, Novara, 2014.

[25] Si veda V. MOLASCHI, *Le arene deliberative,* cit., 60-61.

[26] Per una descrizione dei singoli modelli elencati, *ibidem*, 62-85. Cfr. altresì D. GIANNETTI, *Modelli e pratiche della democrazia deliberativa*, in G. PASQUINO (a cura di), *Strumenti della democrazia*, Bologna, 2007, 139 ss. e U. ALLEGRETTI (a cura di), *Democrazia partecipativa. Esperienze e prospettive in Italia e in Europa*, Firenze, 2010.

[27] Così V. MOLASCHI*, Le arene deliberative,* cit., 64. Della stessa opinione L. BOBBIO, G. POMATTO*, op. cit*., 49.

della cittadinanza potenzialmente interessata da un certo cambiamento e, al termine dello scambio tra cittadini e scienziati, consentono di individuare posizioni condivise sull'utilizzo della tecnologia o della scoperta oggetto della discussione. Vi sono, tuttavia, anche casi in cui l'esito dei lavori non è legato al raggiungimento di un consenso e, pertanto, si parla non più di *consensus conferences* ma di *citizen conferences* o *citizen assemblies*[28]. È questo il caso della *Global citizens' assembly* convocata dal network australiano.

I lavori di queste conferenze sono molto strutturati e, solitamente, articolati in tre momenti: nella prima fase, i cittadini coinvolti ricevono le informazioni necessarie per l'inquadramento dei termini della questione; segue la discussione vera e propria tra laici ed esperti, che devono essere esponenti di diverse tesi e posizioni; da ultimo, l'assemblea produce delle raccomandazioni che vengono sottoposte ai *policy makers* e rese disponibili per l'opinione pubblica.

Questo tipo di arena deliberativa è già stato più volte utilizzato per la discussione di tematiche connesse alla genetica: Ogm, terapie geniche, test genetici, mappatura del genoma umano e così via[29]. Tuttavia, a differenza dell'assemblea globale convocata lo scorso anno, si è sempre trattato di iniziative di dimensione nazionale o sub-statale.

Una delle prime sperimentazioni italiane di *consensus conference* sulla modificazione genetica si è svolta a livello regionale, in Lombardia, nel 2004. In tale occasione, due *panel* di cittadini lombardi, estratti in maniera casuale, sono stati chiamati a discutere con diversi esperti la questione della sperimentazione in campo aperto degli OGM. La conferenza, organizzata dalla Fondazione Bassetti e dall'Istituto di Ricerca della Regione Lombardia, ha portato alla redazione da parte dei cittadini di una serie di raccomandazioni da trasmettere al governo della regione. Tuttavia, trattandosi di una procedura sperimentale, i risultati ottenuti non hanno realmente inciso sulle scelte regionali circa il rilascio di prodotti geneticamente modificati nell'ambiente[30].

Rimanendo nell'ambito di procedure partecipative utilizzate in relazione a questioni scientificamente dibattute o tecnicamente complesse, uno degli ordinamenti più interessanti a cui guardare è, senza dubbio, quello francese. In Francia, infatti, sono stati sperimentati sia il c.d. *débat public* sia gli "Stati generali della bioetica". Il primo è una procedura cui sono sottoposte le decisioni relative all'avvio di alcune categorie di grandi opere, la quale prevede un'importante fase di divulgazione di informazioni al pubblico e, a seguire, un periodo di 4 mesi dedicato al dibattito aperto a tutta la popolazione. All'esito della procedura viene redatto un bilancio che deve essere preso in considerazione dall'ente responsabile della realizzazione dell'opera[31]. Il modello del *débat public* ha ispirato il legislatore italiano nella stesura dell'art. 22 del Codice dei contratti pubblici per le grandi opere infrastrutturali[32], a

[28] Ciò a dimostrazione del fatto che i modelli ideali proposti sono spesso applicati non in maniera pedissequa ma riadattati a circostanze e a obiettivi contingenti (così L. PELLIZZONI, *op. cit*., 150).

[29] Cfr. I report del Loka Institute for Science & Technology of, by & for the people: http://loka.org/TrackingConsensus.html.

[30] Il Policy paper Democrazia partecipativa e legislazione regionale di Eupolis Lombardia, pubblicato nel giugno 2014 e disponibile sul portale: www.polis.lombardia.it .

[31] A riguardo cfr. V. MOLASCHI, *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico,* 478 ss. Cfr. altresì G. PEPE, *Il modello della democrazia partecipativa tra aspetti teorici e profili applicativi un'analisi comparata, cit*., 74 ss e S. CASSESE, *La partecipazione dei privati alle decisioni pubbliche. Saggio di diritto comparato*, in *Rivista trimestrale di diritto pubblico*, 1, 2007, 19 ss.

[32] Cfr. d.lgs. 18 aprile 2016, n. 50, come modificato dal d.lgs. 19 aprile 2017, n. 56.

mente del quale le grandi opere «aventi impatto sull'ambiente, sulle città e sull'assetto del territorio» sono sottoposte alla procedura del dibattito pubblico[33].

Gli Stati generali, invece, consistono in una serie di consultazioni e dibattiti pubblici ad ampia partecipazione, suddivisi in vari *panel* organizzati a livello regionale, che precedono la realizzazione della revisione della legge sulla bioetica[34]. Al termine dei lavori, il *Comité Consultatif National d'Ethique* ha il compito di elaborare un report riassuntivo dei risultati emersi, il quale viene trasmesso al parlamento perché sia fatto oggetto di discussione in sede legislativa[35]. Questo secondo paradigma appare, *ratione materiae*, particolarmente interessante: infatti, tra le questioni bioetiche che hanno definito l'ambito di dibattito degli Stati generali del 2018 si annoverano gli screening genetici e la medicina genomica[36].

All'interno delle eterogenee prassi menzionate, si possono individuare alcune caratteristiche trasversali che particolarmente si confanno alla deliberazione sull'*editing* genetico: innanzitutto, si tratta di luoghi e procedure che consentono di informare, oltre che consultare e acquisire l'opinione della maggior parte degli individui destinatari di una determinata decisione ovvero di un campione il più possibile rappresentativo di costoro; in secondo luogo, «l'effetto della partecipazione non è quello di trasferire il potere decisionale finale in capo ai partecipanti»[37], né quello di vincolare in maniera diretta il legislatore, bensì di aprire un dibattito pubblico e consentire uno scambio di argomenti che trovino sintesi in un prodotto finale (sia esso un report, un bilancio o delle raccomandazioni).

Da un lato, che l'esito della deliberazione non sia vincolante ma al più svolga «una *moral suasion* idonea a condizionare, in via di fatto, la formulazione delle scelte pubbliche»[38], può vedersi come elemento di debolezza delle arene partecipative. Per contro, questa caratteristica contribuisce a mitigare l'obiezione secondo cui sarebbe inadeguato affidare a persone comuni (ossia non scienziati) decisioni basate su acquisizioni e conoscenze complesse[39]. Inoltre, diversamente da quanto accade con uno strumento più incisivo come il *referendum* popolare[40], un'eventuale bocciatura da parte del pubblico di una certa tecnologia non chiude la questione a lungo termine. Al contrario, considerata la natura argomentativa della deliberazione, essa contribuisce a mettere in luce le condizioni alle quali è possibile proseguire.

[33] Sul punto, ampiamente, V. MOLASCHI, *Le arene deliberative, cit*., 258 ss.

[34] Sugli Stati Generali tenutisi nel 2018, cfr. L. GAFFURI, E. PULICE, *Francia - Stati Generali di Bioetica: pubblicato il rapporto di sintesi del Comité consultatif national d'éthique*, in www.biodiritto.org, 5 giugno 2018.

[35] Cfr. A. BLASIMME, *op. cit.,* 1067-1068.

[36] L. GAFFURI, E. PULICE, *op. cit*.

[37] R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa*, *cit*., 4.

[38] G. PEPE, *La democrazia partecipativa ambientale tra ordinamenti sovranazionali ed ordinamento italiano*, in *Federalismi.it*, 2, 2020, 183.

[39] «Should leaders listen […] to the public, some of whom may be convinced their last Whopper contained a Frankenfood patty because an Instagram influencer told them so?» (N. CURATO, S. NIEMEYER, *op. cit*., 2).

[40] Si pensi al caso del referendum abrogativo del 1987 che ha, di fatto, condotto alla fine dello sfruttamento dell'energia nucleare sul territorio italiano. Un secondo referendum, nel 2011, ha escluso la possibilità che si realizzasse nel nostro pianeta un nuovo programma nucleare.

3. La manipolazione genetica come ambito privilegiato per la partecipazione pubblica

Per comprendere se e in che misura il coinvolgimento del pubblico possa e debba giocare un ruolo di rilievo nelle decisioni relative alla regolamentazione del *gene editing*, occorre tenere in considerazione alcuni elementi peculiari delle tecnologie in questione: la connessione con la materia ambientale, l'ampiezza spazio-temporale degli effetti che dalla loro applicazione possono derivare e, naturalmente, la delicatezza etica delle questioni sollevate[41].

Quest'ultimo aspetto, insieme alla divisività del tema nonché alla varietà e rilevanza degli interessi coinvolti, ha fatto delle scelte in materia di organismi geneticamente modificati «*the quintessential type of decision that the public has an explicitly stated desire to participate in*»[42]. Un desiderio che, già comprensibile in relazione all'alterazione genetica di prodotti destinati all'agricoltura – che impatta sull'alimentazione, l'ambiente e l'economia –, è destinato a crescere relativamente all'utilizzo di tecniche che permettono di intervenire sul DNA animale e umano, considerato il maggiore impatto che esse possono avere sulla vita e sul futuro delle persone.

Secondo alcuni, questa richiesta di coinvolgimento dovrebbe trovare tutela in quanto vero e proprio diritto riconosciuto dal diritto internazionale[43], *"the human right to science"*[44]. Esso andrebbe concepito non solo come diritto di poter trarre benefici dal progresso scientifico, bensì anche come diritto ad «una reale partecipazione alla vita scientifica, partecipazione che potrebbe essere intesa anche come il diritto ad essere consultati nel momento della definizione delle attività di ricerca»[45].

Muovendo dalla dimensione dell'interesse dei singoli alla partecipazione, all'orizzonte più ampio del vantaggio per il bene comune, occorre ricordare e fare tesoro della lezione di Asilomar. Come è noto, a metà degli anni '70, Asilomar State Beach fu sede di un'importantissima conferenza sui rischi del DNA ricombinante che coinvolse biologi, medici e giuristi, e che condusse alla elaborazione di una serie di linee guida sull'utilizzo "sicuro" delle tecniche di modificazione genetica e di produzione degli OGM. Sebbene l'iniziativa ebbe il pregio di dimostrare il senso di responsabilità degli scienziati coinvolti, i quali parteciparono allo scopo di individuare dei limiti cui sottoporsi volontariamente nello svolgimento delle proprie attività, essa è stata duramente criticata per aver escluso dal dibattito esponenti "laici" della società civile[46]. Il che, per un verso, ha impedito che insieme alla valutazione dei rischi scientificamente misurabili trovassero il giusto spazio anche argomenti di natura etica e va-

[41] Il che vale soprattutto per la loro applicazione all'uomo: sia sufficiente ricordare che la Dichiarazione universale sul genoma umano e i diritti umani, adottata l'11 novembre 1997 dalla 29 sessione della Conferenza Generale dell'UNESCO, parla del genoma umano come di «patrimonio dell'umanità» (art. 1).

[42] T. ETTY, *Biotechnology*, in *The Yearbook of European Environmental Law*, 5, 2005, 314.

[43] Cfr. in particolare l'art. 27(1) della Dichiarazione Universale del 1948 («Ogni individuo ha diritto di prendere parte liberamente alla vita culturale della comunità, di godere delle arti e di partecipare al progresso scientifico ed ai suoi benefici), nonché l'art. 15(1) del Patto internazionale sui diritti economici sociali e culturali del 1966 («Gli Stati parti del presente Patto riconoscono il diritto di ogni individuo: a) a partecipare alla vita culturale; b) a godere dei benefici del progresso scientifico e delle sue applicazioni; c) a godere della tutela degli interessi morali e materiali scaturenti da qualunque produzione scientifica, letteraria o artistica di cui egli sia l'autore»).

[44] Così M. MANCISIDOR, Is there such a Thing as a human right to science in internatonal law?, in *ESIL Reflections*, 4, 2015.

[45] C. FLAMIGNI, *Sul consenso sociale informato*, in *Biolaw Journal*, 2, 2017, 202.

[46] S. JASANOFF, J.B. HURLBUT, K. SAHA, *CRISPR democracy: gene editing and the need for inclusive deliberation*, in *Issues in Science and Technology*, 32, 1, 2015.

loriale, necessari per stabilire «what forms of progress are culturally and morally acceptable»[47]. Per altro verso, è lecito chiedersi se le profonde divisioni e contestazioni che ancora oggi caratterizzano il dibatto sugli OGM si sarebbero potute attenuare laddove, sin dai suoi esordi, il pubblico fosse stato debitamente informato e consultato[48]. Proprio la lezione di Asilomar ha indotto autorevolissima dottrina ad auspicare che future deliberazioni sul *gene editing* (e in particolare sul CRISPR) muovano da un ripensamento del rapporto tra scienza e democrazia[49]. Da tale ripensamento lo stesso sviluppo tecnologico potrebbe trarre beneficio, laddove ne derivasse una minore avversione da parte del pubblico che – quando non adeguatamente informato – tende a guardare con timore al progresso.

Un secondo elemento che fa dell'ingegneria genetica un ambito d'elezione per il deliberativismo sono i suoi punti di contatto con la materia ambientale. Il dibattito sulla democrazia ambientale[50] e sulla partecipazione nei processi di *decision-making* che riguardano l'ambiente è terreno da tempo arato ed ha condotto ad importanti risultati, in particolare sul piano del diritto internazionale[51]: basti pensare all'approvazione della Convenzione di Aarhus sull'accesso alle informazioni, la partecipazione dei cittadini e l'accesso alla giustizia in materia ambientale[52], implementata a livello europeo dal Regolamento CE n. 1367/2006. Non sempre le forme in cui si realizza la partecipazione democratica alle decisioni ambientali ricalcano gli schemi propri della democrazia partecipativa ma, anche grazie alla Convenzione di Aarhus[53], l'ambiente è indubbiamente considerato «un contesto atto ad accogliere le nuove frontiere della partecipazione che la democrazia deliberativa esprime»[54]. Infatti, questo settore, «rivelando insofferenza per la tradizionale partecipazione procedimentale, ha, nel corso degli anni, preteso forme di più ampio dialogo tra istituzioni e consociati in vista dell'acquisizione e del

[47] *Ibidem*, 3.
[48] Similmente, *ibidem*, 5.
[49] *Ibidem*, 3.
[50] A riguardo cfr. *ex multis* M. MASON, *Environmental Democracy: A Contextual Approach*, Londra, 1999; C. PITEA, *Diritto internazionale e democrazia ambientale*, Napoli, 2013; G. PAROLA, *Environmental Democracy at the Global Level: Rights and Duties for a new Citizenship*, Londra, 2013; G. PEPE, *La democrazia partecipativa ambientale tra ordinamenti sovranazionali ed ordinamento italiano,* cit., 179 ss.
[51] Il diritto internazionale ha avuto, altresì, il merito di sottolineare l'importanza della partecipazione del pubblico nella promozione dello Sviluppo Sostenibile (cfr. par. 43 della Risoluzione delle Nazioni Unite A/RES/66/288 "The Future We Want"). Ciò emerge chiaramente anche all'interno dell'Agenda 2030 per lo sviluppo sostenibile e, in particolare nel Goal 16. Sul punto cfr. G. RAGONE, *The GMO Authorization Procedure in EU: Inclusivity, Access to Justice and Participation in Decision-Making, in Diritto Pubblico Europeo Rassegna online*, 2, 2019, 206 ss.
[52] Sul punto, S.T. MCALLISTER, *The Convention on Access to Information, Public Participation in Decision-Making, and Access to Justice in Environmental Matters*, in *Colorado Journal of International Environmental Law and Policy*, 1999, 187 ss.; M. LEE, C. ABBOT, *The usual suspects? Public participation under the Aarhus Convention*, in *Modern Law Review*, 66, 2003, 80 ss.; M. PALLEMAERTS (a cura di), *The Aarhus Convention at Ten: interactions and tensions between conventional international law and EU environmental law*, in *Europa law publishing*, 2011.
[53] Ed in particolare al suo Secondo Pilastro.
[54] V. MOLASCHI, *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico,* cit., 464.

bilanciamento della moltitudine di interessi in rilievo»[55] e, come si è visto, proprio nella direzione di questo dialogo vanno le arene deliberative descritte al paragrafo precedente.

Che la coltivazione degli OGM rientri in questo contesto è fuori discussione e, in anni recenti, il Tribunale di prima istanza dell'Unione Europea[56] ha stabilito che anche le decisioni relative alla loro commercializzazione possono essere considerate questioni di diritto ambientale[57]. Ne discende che, all'interno dell'Unione, i c.d. *participation rights* riconosciuti dalla Convenzione di Aarhus e dall'omonimo Regolamento vengono in rilievo anche nei processi di autorizzazione all'immissione in commercio degli OGM. È plausibile attendersi che ulteriori estensioni dell'ambito di applicazione dei diritti in questione si verifichino in futuro al progredire delle tecniche di ingegneria genetica. In qualche misura ciò è già avvenuto, sempre per via pretoria, quando nel 2018 la Corte di Giustizia dell'Unione Europea[58] ha stabilito che, per quanto concerne il diritto dell'Unione, sono assoggettati alla disciplina riservata agli OGM sia i prodotti ottenuti tramite transgenesi (ossia inserendo nell'organismo DNA estraneo), sia quelli realizzati invece mediante mutagenesi. La Corte ha, inoltre, chiarito che gli obblighi relativi agli OGM si rivolgono anche agli organismi ottenuti mediante tecniche di mutagenesi emerse successivamente all'adozione della normativa di riferimento, come nel caso del CRISPR.

Se l'ambiente è terreno fertile per il deliberativismo è anche perché, in questo contesto più che in altri, occorre «prendere atto che [il diritto] non è più solo il prodotto di una sovranità popolare istituzionalizzata ma è anche il risultato di una sovranità popolare sempre più non istituzionalizzata»[59]. Infatti, mentre dalle tradizionali istituzioni rappresentative dipendono decisioni che incidono sui territori da cui esse traggono legittimazione, le scelte che riguardano l'ambiente hanno generalmente portata più ampia, trascendono i confini territoriali e si ripercuotono su una popolazione che non necessariamente appartiene ad un medesimo ordinamento.

I nessi tra deliberativismo e globalizzazione sono stati messi in luce da diversi studiosi. Alcuni sottolineano come i processi di globalizzazione, contribuendo ad alimentare la crisi della istituzioni rappresentative tradizionali[60], abbiano come effetto consequenziale quello di favorire il diffondersi delle teorie deliberativiste. Altri, osservando il passaggio da *government* a *governance* favorito dalla globalizzazione, suggeriscono che – in assenza di istituzioni rappresentative di dimensioni globali – siano proprio le arene deliberative la sede ideale in cui discutere decisioni che possono avere portata globale[61]. È questo il caso della regolamentazione di tecnologie impattanti come il *gene editing*, incluse quelle applicazioni che non possono essere ricondotte alla materia ambientale, ed in particolare a

[55] G. Pepe, *La democrazia partecipativa ambientale tra ordinamenti sovranazionali ed ordinamento italiano, cit.*, 183.

[56] *TestBiotech et al. v. Commissione* (T-177/13), 15 dicembre 2016.

[57] Cfr. G. Ragone, *Il delicato ruolo del giudice tra valutazioni scientifiche controverse e scelte politico-discrezionali in materia di OGM*, in *DPCE online*, 4, 2016, 252.

[58] Confédération paysanne et al. (C-528/16), 25 luglio 2018. Cfr. M.C. Errigo, *Diritto e OGM. Una storia complicata*, in *BioLaw Journal*, 1, 2020, 304.

[59] R. Bifulco, *Democrazia deliberativa e democrazia partecipativa,* cit., 9.

[60] Cfr. ad esempio G. Pepe, *Il modello della democrazia partecipativa tra aspetti teorici e profili applicativi un'analisi comparata,* cit., 13 ss.

[61] Così J.S. Dryzek, A. Bächtiger, K. Milewicz, *Toward a Deliberative Global Citizens' Assembly,* cit., 33 ss.; J.S. Dryzek et al., *Global citizen deliberation on genome editing,* cit., 1435 ss.

ISSN 2284-4503

quelle riferite al genoma umano: «the human genome is not the property of any particular culture, nation or region; […] It belongs equally to every member of our species»[62]. Questa consapevolezza, insieme alla imponderabilità dell'ampiezza degli effetti nel tempo degli interventi sul DNA umano (soprattutto se condotti sulla linea germinale), ha condotto ad affermare che «decisions about how far we should go in tinkering with it have to be accountable to humanity as a hole»[63].
Ma come immaginare di poter consultare cittadini di tutto il mondo ed instaurare con loro un dialogo sui limiti entro i quali possono spingersi tecnologie tanto complesse? La *Global citizens' assembly* di Canberra costituisce un primo tentativo di raccogliere questa sfida.

4. Il caso della "Global citizens' assembly on genome editing"

La convocazione di un'assemblea cittadina globale sulla manipolazione genomica richiede di affrontare aspetti problematici e limiti attuativi di non facile superamento[64]. In particolare: in che modo selezionare i partecipanti? Come organizzare e a chi far dirigere lo svolgimento dei lavori? E ancora: quale impatto ci si può attendere da un simile esperimento?
In generale, l'aspetto della selezione del pubblico è una delle questioni più spinose[65] giacché, oltre ad incidere significativamente sulla buona riuscita dell'iniziativa, rappresenta un chiaro paradosso: con la democrazia partecipativa «si ambisce a includere tutti, ma – di fatto – si riesce a concretamente a coinvolgere solo qualcuno»[66].
I metodi solitamente utilizzati nell'ambito delle arene deliberative sono riconducibili a tre archetipi, i quali possono essere applicati in maniera mista: il metodo della porta aperta, il microcosmo e il mini-pubblico[67]. Il primo meccanismo consiste in un'autoselezione: ogni cittadino che lo voglia e che sia informato sull'esistenza di una determinata arena può prendervi parte sino al raggiungimento di un numero congruo. Il secondo metodo presuppone che un organizzatore esterno al *forum* proceda ad una selezione dei partecipanti, cercando di creare una rappresentanza il più possibile fedele di tutti coloro che hanno un interesse specifico alla deliberazione. In questo caso i partecipanti finiscono con l'essere portatori di una visione precostituita e danno luogo ad una discussione "calda"[68]: ciò costituisce un vantaggio sotto il profilo della conoscenza del tema da parte dei cittadini ma va a detrimento della possibilità che lo scambio argomentativo sia genuino e fruttuoso. Da ultimo, l'archetipo dei

[62] S. JASANOFF, J.B. HURLBUT, K. SAHA, *op. cit.*, 2.
[63] *Ibidem*, 2.
[64] Più in generale, sulle criticità realizzative delle assemblee deliberative cfr. E. CAROLAN, *op. cit.*, passim. L'Autore, riferendosi alla fase della deliberazione vera e propria, sottolinea come affatto neutre siano le scelte relative a «how to frame an issue; in what order it should be approached; from what disciplinary perspectives; using what kinds of 'evidence' and 'experts'; and subject to what limitations; shape how the process develops». Tutti questi elementi «inevitably privilege particular kinds of input and require difficult trade-offs between abstract deliberative goods like expertise, independence, experience, impartiality, participation, openness and evidential scrutiny».
[65] Così anche L. PELLIZZONI, *op. cit.*, 150
[66] L. BOBBIO, G. POMATTO, *op. cit.*, 52.
[67] Cfr. R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa*, *cit.*, 5; L. BOBBIO, G. POMATTO, *op. cit.*, 52-53.
[68] *Ibidem*, 58.

mini-publics[69] comporta la selezione casuale di un campione della popolazione, eventualmente stratificato in quote sociodemografiche (per genere, età, provenienza e così via): «la scommessa è che qualsiasi cittadino, messo in condizioni di interloquire con gli altri e di assumere le informazioni necessarie, sia in grado di esprimere posizioni puntuali su qualsiasi problema pubblico e costruire, insieme agli altri, soluzioni intelligenti»[70]. Questo tipo di selezione porta ad un dibattito tendenzialmente libero da interessi partigiani[71] ed è il più comune nell'ambito delle *consensus conferences* e *citizens' assemblies*. Non fa eccezione la *Global Citizens' Assembly* australiana.

In questo caso l'estrazione mira a selezionare un numero di partecipanti compreso tra 24 e 100 (dipenderà dall'ampiezza dei finanziamenti a disposizione), individuando persone che provengano da tutti i continenti del pianeta e che riflettano il più possibile la comunità globale quanto a età, genere, livello di istruzione, etnia e residenza. Se i fondi lo consentiranno, l'assemblea globale sarà preceduta da *forum* organizzati a livello nazionale e, in tal caso, è possibile che alcuni dei partecipanti vengano scelti tra coloro che prenderanno parte alle arene preparative.

Una volta costituito il *panel* dei partecipanti, i lavori saranno organizzati e guidati dal centro *Deliberative Democracy and Global Governance* dell'Università di Canberra, anche se la sede dell'assemblea non sarà necessariamente in Australia. È piuttosto usuale che questo genere di conferenze sia seguito passo passo da professionisti della deliberazione, in grado di garantire il corretto svolgimento di tutte le fasi dei lavori.

Nel caso di specie, il programma prevede cinque giornate lavorative. La prima sarà dedicata alla presentazione delle regole di funzionamento della conferenza e alla reciproca conoscenza dei partecipanti. I successivi tre giorni saranno destinati alla trattazione rispettivamente della modificazione genetica di piante, animali ed essere umani. La discussione sarà condotta in gruppi, alla presenza di eticisti, giuristi e scienziati, ai quali sarà chiesto di condividere le proprie conoscenze. Nel quinto e ultimo giorno, i partecipanti saranno chiamati a redigere un documento contenente raccomandazioni pratiche rivolte ai decisori politici, nonché alla comunità scientifica, identificando le questioni etiche ritenute più problematiche e i principi regolatori che si considerano più adeguati alla materia. Il documento di sintesi così ottenuto sarà presentato, oltre che ad autorità e legislatori nazionali, ad attori di rilievo globale (quali il Segretario Generale delle Nazioni Unite e i direttori generali dell'OMS e della FAO).

Sebbene sprovvisto di valore giuridico e destinato ad essere successivamente modificato per la necessità di riaggiornare il dibattito al progredire della ricerca in ambito genetico[72], il report finale avrà lo scopo di offrire ai decisori un punto di vista che difficilmente potrebbe essere acquisito per altre vie: quello dei cittadini – della comunità globale – su cui le decisioni si ripercuoteranno.

[69] Termine utilizzato per la prima volta in A. FUNG, *Recipes for public spheres: Eight institutional design choices and their consequences*, in *Journal of Political Philosophy*, 3, 2003, 338 ss.

[70] L. BOBBIO, G. POMATTO, *op. cit.*, 56-57.

[71] Sui vantaggi e svantaggi delle deliberazioni "fredde", cfr. L. BOBBIO, G. POMATTO, *op. cit.*, 58.

[72] J.S. DRYZEK et al., *Global citizen deliberation on genome editing,* cit., 1437.

5. Conclusioni

La portata innovativa delle tecnologie di ingegneria genetica, applicabili a tutte le forme di vita del pianeta (tra cui l'uomo), impone di riflettere su quali siano i processi decisionali più idonei a incidere su quello che è stato definito «our genomic future»[73]. In questa sede si è scelto di prendere in considerazione l'opportunità di coinvolgere il pubblico attraverso strumenti partecipativi basati sui principi della democrazia deliberativa, focalizzandoci in particolare sul modello offerto dalle c.d. *consensus conferences*. Come si è visto, diversamente dagli istituti di democrazia diretta, «questi spazi di partecipazione e deliberazione […] non hanno, e non possono rivendicare, alcun diretto potere decisionale, ma non per questo sono irrilevanti»[74].

Tra i pregi che sono stati riconosciuti a questo genere di esperienze democratiche, vi è innanzitutto quello di aprire un dialogo tra istituzioni e società civile[75]. Il che può «giocare un ruolo decisivo come fattore di legittimazione (o criticamente, di de-legittimazione) delle decisioni assunte delle istituzioni»[76], arricchendo il tasso di democraticità dei nostri ordinamenti[77]. Il dialogo col pubblico nella fase che precede il momento decisionale vero e proprio può, inoltre, contribuire all'assunzione di scelte migliori: i *decision makers* possono infatti basarsi su considerazioni ulteriori rispetto a quelle che vengono loro offerte dagli esperti della materia.

In sintesi, se le decisioni relative a queste tecnologie fossero anticipate da una pubblica discussione, esse godrebbero di una maggiore legittimazione[78] e vi sarebbe modo di far emergere – e correggere per tempo – le criticità percepibili dal pubblico. Naturalmente, come si è già sottolineato, non è automatico che all'esito della deliberazione corrisponda necessariamente un impatto migliorativo sulla decisione finale: questo sia per la possibile inesperienza dei partecipanti, sia per la mancanza di vincolatività delle loro raccomandazioni. In un certo senso, il rapporto tra i cittadini che partecipano alla deliberazione e i decisori finali è paragonabile a quello che intercorre tra gli *amicus curiae* e le corti: il loro parere può essere ignorato ma, al contempo, il loro punto di vista entra nell'orizzonte conoscitivo del soggetto cui spetta il diritto all'ultima parola.

Un ulteriore effetto benefico della istituzione di arene deliberative su questioni scientifiche e tecnologiche emergenti è quello di ridurre il populismo anti-scientitista, accrescendo la fiducia del pubblico su come i *policy-makers* si avvalgono di pareri esperti[79]. Come si è potuto osservare nell'ambito della gestione dell'emergenza globale conseguita alla pandemia da Covid-19, per un verso il ruolo degli esperti (e dei comitati dotati delle opportune competenze tecnico-scientifiche) è fondamentale per supportare i processi decisionali in determinati settori; per contro, le divisioni all'interno della comu-

[73] A. GREENFIELD, *Our genomic future*, in *News Scientist*, 241, 2019, 24-25.

[74] A. FLORIDIA, *Democrazia partecipativa e democrazia deliberativa: una risposta plausibile alla "crisi della democrazia"?,* cit., 3.

[75] J.S. DRYZEK et al., *Global citizen deliberation on genome editing,* cit., 1436.

[76] *Ibidem*, 3.

[77] R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa*, cit., 7.

[78] La più forte legittimazione delle decisioni precedute da consultazione pubblica impone di riflettere, in seconda battuta, sulle modalità e sul grado di pervasività che il sindacato giurisdizionale può adottare su di esse. Circa i modelli di controllo da parte delle corti sulle procedure di partecipazione, cfr. S. CASSESE*, op. cit*., 18 ss.

[79] Cfr. V. MOLASCHI, *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico, cit.*, 469; Cfr. A. BLASIMME, *op. cit*., 1068.

nità scientifica, l'opacità sui criteri con cui si sceglie di fare affidamento su una determinata visione e l'instabilità delle acquisizioni scientifiche, soggette a continue revisioni e rivalutazioni nel tempo, possono essere causa di scetticismo da parte dei consociati circa la bontà delle decisioni assunte.
Permettere ad una parte della popolazione interessata da una determinata innovazione di porre domande e interloquire con scienziati ed esperti, oltre a costituire un interessante esempio di co-produzione tra scienza e società[80], aiuta la creazione di un "consenso sociale informato"[81]. Questo principio, che «bussa alla porta della bioetica da tempo»[82] e che esprime un rapporto ideale tra cittadini e ricerca scientifica, «può svolgere una funzione di garanzia e di promozione della scienza in un periodo nel quale i suoi quotidiani progressi suscitano speranze e preoccupazioni crescenti»[83].
Volgendo a conclusioni, sembra potersi affermare che il potenziale insito nella realizzazione di luoghi di partecipazione pubblica a decisioni *science and technology based*, come l'assemblea cittadina globale annunciata dal *Centre for Deliberative Democracy and Global Governance*, sia quello di muovere dei passi verso il «superamento della crisi di legittimazione di cui soffrono le istituzioni scientifiche e politiche»[84]. Ciò suggerisce la necessità di proseguire il dibattito sul deliberativismo e sulle sue concrete modalità attuative per contribuire a superarne i limiti che nel corso della trattazione si è cercato di rilevare.

[80] Cfr. M. TALLACCHINI, *op. cit*., passim.
[81] Sul concetto, cfr. C. FLAMIGNI, *op. cit*., passim.
[82] *Ibidem*, 202.
[83] *Ibidem*, 201.
[84] V. MOLASCHI, *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico,* cit., 470.

Special issue

Gene editing decisions and democratic participation: A privileged area for the application of the principles of deliberative democracy?

Giada Ragone *

ABSTRACT: The frontiers reached by gene editing and their applications for animals, plants and humans raise numerous tricky issues, posing new and renewed challenges for constitutional law scholars. This paper focuses on the role that the public can play in decision-making processes aimed at regulating gene-editing technologies. Thus, it reflects on the opportunity to develop and enhance deliberative arenas, which guarantee a fuller participation of citizens in legislative choices that touch upon controversial scientific issues of general interest, in compliance with the principles of deliberative democracy.

KEYWORDS: deliberative democracy; participatory democracy; deliberative arenas; gene editing; public participation

SUMMARY: 1. Introduction – 2. Deliberativism and participatory democracy: A theoretical framework and models to look at – 3. Genetic manipulation as a privileged field for public participation – 4. The case of the Global Citizens' Assembly on Genome Editing – 5. Conclusions.

1. Introduction

In 2020, the Centre for Deliberative Democracy and Global Governance at the University of Canberra (Australia) and an international network of researchers announced the convening of the first Global Citizens' Assembly on Genome Editing.[1] The initiative consists of creating a forum where citizens from all over the world can discuss, exchange opinions, and receive information on the developments and implications of the most advanced genetic engineering techniques, which now make it possible to alter the genetic sequence of any organism (plant, animal or human) "to

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[1] See N. CURATO, S. NIEMEYER, *Why we need a global citizens' assembly on gene editing*, in *TheConversation.com* , 17 settembre 2020; J.S. DRYZEK et al., *Global citizen deliberation on genome editing, in American Association for the Advancement of Science*, 369, 6510, 18 September 2020, 1435-1437; J.S. DRYZEK, A. BÄCHTIGER, K. MILEWICZ, *Toward a Deliberative Global Citizens' Assembly,* in *Global Policy*, 1, 2011, 33-42; J. CHADWICK, *Plumbers and teachers are invited to share their views on designer babies and genetically enhanced potatoes as part of a citizens' assembly on 're-engineering' the human species*, in *Daily Mail Online*, 18 September 2020. The official website of the "Global Citizens' Assembly on Genome Editing. Connecting citizens, science and global governance" project is: https://www.globalca.org/.

produce favourable characteristics or remove unwanted ones".[2] At the end of the deliberative phase, the assembly is asked to produce recommendations ("moral and political regulation"[3]) that can be taken into account by those who are responsible for regulating these technologies at all levels.

For decades, the legal debate on genetic manipulation has focused mainly on the production of GMO foods – that is, agricultural products for human and animal consumption, the genetic material of which is modified in a way that differs from what occurs in nature. However, the DNA alteration techniques that have been developed over the years not only concern the gene modification of seeds and plant varieties, but also involve all life forms on the planet, including humans.[4]

One of the most promising and versatile technologies in this field is the CRISPR-Cas,[5] which makes it possible to cut and replace genome sequences in a relatively simple way, applying a system that is naturally present in bacteria and other single-celled organisms. Genetic 'correction' can take place either in the germ line of an organism (i.e. before cell differentiation occurs) or later in its development, in the somatic line. When applied to germ cells, the modification not only affects the organism in which it is carried out – it will be passed on to all organisms that later receive its genetic heritage. Thus, manipulation of the germline entails the possibility that unforeseeable side-effects of the alteration will turn into permanent genetic mutations.[6]

Despite these shadows, there is no doubt that the potentials of CRISPR are vast and promising. They range from the improvement of food, to medical applications of enormous scope (e.g. the possibility of stopping the spread of diseases, such as malaria, by intervening in the DNA of insects that carry it,[7] helping in the fight against cancer, and curing rare genetic diseases), and even to altering human embryos produced in vitro to remove from the genetic make-up of unborn children any genes considered to be decisive in the development of diseases or undesirable characteristics. In 2018, CRISPR was used to give birth to the first two genetically modified human beings in a controversial and opaque experiment carried out by the Chinese researcher Jiankui He. According to the scientist, the clinical trial he conducted involved couples of would-be parents made up of HIV-positive fathers and led to the production of embryos resistant to the virus in question by deactivating the gene that usu-

[2] N. CURATO, S. NIEMEYER, *op. cit*., 1.

[3] J.S. DRYZEK et al., *Global citizen deliberation on genome editing*, *cit*., 1435.

[4] The first famous attempt to genetically modify human embryos dates back to 2015, which was followed by the convening of an international summit of scholars (the International Summit on Human Gene Editing) in Washington, conceived as "an initial attempt to keep the discussion about human genome editing thematically broad and open to input from a variety of stakeholders" (A. BLASIMME, *Why Include the Public in Genome Editing Governance Deliberation?*, in *AMA Journal of Ethics*, 21, 12, 2019, 1067).

[5] Acronym for *Clustered Regularly Interspaced Short Palindromic Repeats-Cas*. On this technique see https://bit.ly/3gBT8b7 .

[6] Therefore, as recalled in S. BONOMELLI, *Gene editing embrionale: il vaso di pandora è stato scoperchiato? Riflessioni a margine del caso di Jiankui He*, in *BioLaw Journal*, 3, 2019, 72, both the Oviedo Convention and the Universal Declaration on the Human Genome and Human Rights UNESCO absolutely prohibit genetic interventions on the germ-line.

[7] See M. ANNONI, T. PIEVANI, *What is wrong in extinguishing a species? Charting the Ethical Challenges of using Gene-Drive Technologies to eradicate* A. gambiae *vector populations* in this Special Issue.

ally allows contagion in humans.[8] The genetic editing operation was therefore intended to prevent the children born from the experiment from contracting HIV in the future.[9]

It is quite clear that the advances made in gene editing and the horizons towards which such technologies are moving raise numerous delicate questions, not only on the ethical and scientific levels, but also on the legal one, posing new and renewed challenges for constitutional law scholars.[10] In this paper, we have chosen to reflect, in particular, on the role that the general public can play in decision-making processes aimed at regulating these technologies. In this way, we intend to contribute to the debate on the opportunity to develop and enhance deliberative arenas[11] that, like the one convened by the scholars of the University of Canberra, guarantee a fuller participation of the public in regulatory choices that involve controversial scientific issues of general interest,[12] in accordance with the principles of so-called deliberative democracy.[13]

To this end, we will first define deliberativism and attempt to identify the models of participatory democracy that appear to be the most suitable to be taken into consideration for science- and technology-based decisions. It will be made clear, however, that "the models adopted are very varied, so that it is not possible to refer to a standardised model, but rather to certain guiding principles".[14]

Second, an examination will be made of the characteristics that make the chosen field, namely gene editing, a particularly interesting test bed for the application of deliberativist theories.

Finally, before turning to conclusions, the merits and limitations of the Global Citizens' Assembly on Gene Editing will be highlighted, with particular emphasis on the choice of conducting a global experiment.

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[8] On this experiment see S. BONOMELLI, *op. cit.,* 67 ss.

[9] However, the experiment seems to have largely failed. See D. PERRIN – G. BURGIO, *China's failed gene-edited baby experiment proves we're not ready for human embryo modification*, in *TheConversation.com*, 9 December 2019.

[10] On the "interactions between the two 'codes' (genetics and law) that make up the 'palimpsest of life' in the process of 'bioconstitutional' reconfiguration of rights" (M. TALLACCHINI, *Scienza e diritto. Prospettive di co-produzione*, in *Rivista di filosofia del diritto*, 2, 2012, 317) see S. JASANOFF (ed.), *Reframing Rights. Bioconstitutionalism in the Genetic Age, Cambridge*, 2011.

[11] See L. BOBBIO, *Le arene deliberative*, in *Rivista Italiana di Politiche Pubbliche*, 3, 2002, 5 ss. and more recently V. MOLASCHI, *Le arene deliberative. Contributo allo studio delle nuove forme di partecipazione nei processi di decisione pubblica*, Napoli, 2018 and ID., *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico*, in *Nuove Autonomie*, 3, 2017, 464. Here, deliberative arenas are referred to as "instruments, inspired by the principles of deliberative democracy, which are increasingly being used in the case of public decisions concerning scientifically and technically complex issues, typically in the fields of health and biomedical research, biotechnology and the environment".

[12] For the definition of a "controversial scientific issue", refer to L. VIOLINI, *Le questioni scientifiche controverse nel procedimento amministrativo*, 1986, Pavia.

[13] On this subject, in the Italian literature, see *inter alia*, U. ALLEGRETTI, *Basi giuridiche della democrazia partecipativa in Italia: alcuni orientamenti*, in *Democrazia e diritto*, 3, 2006, 151 ss. e ID., *Democrazia partecipativa*, in *Enciclopedia del Diritto – Annali IV*, Milano, 2011, 295 ss.; R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa. Relazione al Convegno "La democrazia partecipativa in Italia e in Europa: esperienze e prospettive"* - Firenze, 2-3 aprile 2009, in *Rivista Astrid* e ID., *Democrazia deliberativa e principio di realtà*, in *Federalismi.it*, numero speciale 1, 2017, 1 ss.; A. FLORIDIA, *La democrazia deliberativa. Teorie, processi e sistemi*, Carocci, Roma, 2013 e ID. *Democrazia partecipativa e democrazia deliberativa: una risposta plausibile alla "crisi della democrazia"?,* in www.fondazionefeltrinelli.it, 29 marzo 2019.

[14] U. ALLEGRETTI*, Basi giuridiche della democrazia partecipativa in Italia: alcuni orientamenti, cit.,* 152.

2. Deliberativism and participatory democracy: A theoretical framework and models to look at

Usually, when describing the institutions and mechanisms that characterise our contemporary democracies, recourse is made to the concepts of "representative democracy" and "direct democracy". If we take the Italian Constitution as an example, it outlines a democratic state that entrusts the main decision-making functions to state bodies that *represent* the people and are linked to them through direct or indirect elective mechanisms; at the same time, there are also institutions of so-called direct democracy, such as referendums, popular legislative initiatives and petitions, which allow the electoral body to make decisions that affect the system *without the mediation* of representatives.

The creation of institutions of direct democracy within representative systems responds to the need "to ensure popular participation in decisions that affect the entire community and to bridge the gap between the people and the state apparatus".[15] The institutions or procedures that the doctrine frames as examples of "participatory democracy" and that have their roots in the various theories of "deliberative democracy"[16] tend towards a similar objective, albeit in different ways.[17] It is not by chance that the diffusion of instruments typical of these meanings of democracy is considered, inter alia, a consequence of the crisis of representative democracy.[18]

Two clarifications are necessary before proceeding to a definition. First of all, we agree with the view that "Participatory Democracy and Deliberative Democracy are not alternative forms to representative democracy; rather, they are models or ideas that can indicate certain characteristics and mark the greater or lesser quality of representative democracy itself".[19] The institutions and forms that are generally brought under the umbrella of these concepts can therefore be seen as complementary to the institutions of representative democracy and direct democracy.[20]

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[15] R. BIN, G. PITRUZZELLA, *Diritto costituzionale*, XXI ed., Torino, 2020, 75.

[16] On the possibility that the implementation of forms of participatory democracy offers a credible response to the need for citizens to participate in the life of society and institutions, see U. ALLEGRETTI, *Basi giuridiche della democrazia partecipativa in Italia: alcuni orientamenti, cit.*, 151.

[17] L. BOBBIO, G. POMATTO, *Il coinvolgimento dei cittadini nelle scelte pubbliche*, in *Meridiana: rivista di storia e scienze sociali*, 58, 2007, 46. Contra see G. SCOFFONY, *La démocratie participative dans les États fédérés américains,* in F. ROBBE (ed.), *La démocratie participative*, Paris, 2007, 98, according to which the institutions of direct democracy are also forms of participatory democracy.

[18] See V. MOLASCHI, *Le arene deliberative, cit.,* 18-19; G. PEPE, *Il modello della democrazia partecipativa tra aspetti teorici e profili applicativi un'analisi comparata*, Padova, 2020, 13 ff. and A. FLORIDIA, *Democrazia partecipativa e democrazia deliberativa: una risposta plausibile alla "crisi della democrazia"?, cit*., passim.

[19] A. FLORIDIA, *Democrazia partecipativa e democrazia deliberativa: una risposta plausibile alla "crisi della democrazia"?, cit.,* 2.

[20] Cfr. U. ALLEGRETTI, *Basi giuridiche della democrazia partecipativa in Italia: alcuni orientamenti, cit*., 153; M. SETÄLÄ, *Connecting deliberative mini-publics to representative decision making*, in *European Journal of Political Research*, 56, 2017, 846 ff. An interesting – albeit not without criticism – combination of instruments of direct democracy and participatory democracy has been realised on several occasions in Ireland, where important referendums on ethically sensitive issues have been anticipated by deliberative assemblies: see E. CAROLAN, *Ireland's Citizens' Assembly on Abortion as a Model for Democratic Change? Reflections on Hope, Hype and the Practical Challenges of Sortition*, in *IACL-AIDC Blog*, 28 November 2018; C. O' CINNEIDE, *The Citizens' Assembly Viewed in External Perspective: Useful, but not a Deliberative Deus Ex Machina*, in *IACL-AIDC Blog*, 12 December 2018.

The second clarification concerns the relationship between deliberative and participatory democracy: although there are divergent reconstructions, here we adhere to the approach according to which the deliberativist doctrines[21] constitute the theoretical basis on which the practices attributable to participatory democracy are founded.[22]

It is not easy to offer a single definition of deliberativism. It constitutes a highly varied theoretical background.[23] However, it is possible to trace two characteristics that serve as the lowest common denominator among its various theorisations:[24] first, it is a concept of democracy in which public decision-making processes must include a phase of exchange of opinions and information that is as complete and impartial as possible; second, the discussion (or deliberation) must be inclusive, ideally involving all the subjects concerned by the final decision, or at least a representative sample.

With regard to the forms that participatory democracy can take, many practices have been tried out all over the world, both at national and local levels:[25] suffice it to say that more than a hundred different types of participatory mechanisms have been counted in the literature.[26] Among the best known and most widespread models are citizens' juries, deliberative polls, town meetings, *Planungszellen*, participatory budgets and consensus conferences.[27] The latter, in particular, are relevant to the theme of this paper, since they constitute "a typical example of a participatory process aimed at the democratisation of public decisions, so-called science and technology based".[28] In fact, they put experts on a given scientific-technical issue in dialogue with a sample of the citizenry potentially affected by a certain change and, at the end of the exchange between citizens and scientists, allow the identification of shared positions on the use of the technology or discovery under discussion. However, there are also cases in which the outcome of the work is not linked to the achievement of a consensus; therefore, these conferences should not be considered consensus conferences but rather "citizen conferences" or "citizen assemblies".[29] Such is the case of the Global Citizens' Assembly convened by the Australian network.

[21] The first theorisation of the concept of deliberative democracy can be found in J.M. BESSETTE, *Deliberative Democracy: The Majority Principle in Republican Government*, in R. GOLDWIN, W.A SCHAMBRA (a cura di), *How Democratic is the Constitution?*, Washington, 1980, 102 ss.

[22] R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa*, *cit.,* 2 ss.

[23] *Idem*, 3.

[24] Cfr. V. MOLASCHI, *Le arene deliberative, cit.,* 34 ss. According to Pellizzoni, who speaks of deliberative democracy as the "most significant innovation in democratic debate and practice in recent decades" (L. PELLIZZONI, *Comunità, partecipazione e democrazia deliberativa: un'esperienza italiana*, in *Quaderni di sociologia*, 68, 2015, 149), the key option of the deliberative ideal is the establishment of organised discussion.

[25] For an interesting example of a deliberative arena organised at municipal level see G. BALDUZZI, D. SERVETTI (eds.), *Discutere e agire. Una sperimentazione di democrazia deliberativa a Novara*, Novara, 2014.

[26] See V. MOLASCHI, *Le arene deliberative, cit*., 60-61.

[27] For a description of the individual models listed, *ibidem*, 62-85. See also D. GIANNETTI, *Modelli e pratiche della democrazia deliberativa*, in G. PASQUINO (a cura di), *Strumenti della democrazia*, Bologna, 2007, 139 ss. e U. ALLEGRETTI (a cura di), *Democrazia partecipativa. Esperienze e prospettive in Italia e in Europa*, Firenze, 2010.

[28] See V. MOLASCHI*, Le arene deliberative, cit*., 64. The same opinion is shared by L. BOBBIO, G. POMATTO*, op. cit*., 49.

[29] This demonstrates that the ideal models proposed are often not applied blindly but adapted to circumstances and objectives (L. PELLIZZONI, *op. cit*., 150).

The work of these conferences is highly structured and usually consists of three stages: in the first stage, the citizens involved receive the information needed to frame the terms of the issue; this is followed by the actual discussion between laypeople and experts, who must be exponents of different theses and positions; finally, the assembly produces recommendations that are submitted to policy makers and made available to the public.

This type of deliberative arena has already been used several times to discuss issues related to genetics: GMOs, gene therapies, genetic testing, human genome mapping and so on.[30] However, unlike the global assembly convened last year, these have always been national or sub-state initiatives.

One of the first Italian consensus conference experiments on genetic modification took place at the regional level, in Lombardy, in 2004. On this occasion, two randomly selected panels of Lombardy citizens were asked to discuss the issue of open-field experimentation with GMOs with various experts. The conference, organised by the Bassetti Foundation and the Research Institute of the Lombardy Region, resulted in the citizens drawing up a series of recommendations to be forwarded to the regional government. However, as this was an experimental procedure, the results obtained did not really affect regional decisions on the release of genetically modified products into the environment.[31]

One of the most interesting systems to look at is undoubtedly that of France, where participatory procedures are used in relation to scientifically debated or technically complex questions: this is the case of the so-called *débat public* and the *États généraux de la Bioethique* have been tried out. The former is a procedure to which decisions on the launch of certain categories of major works are subject, which includes an important phase of public information and then a four-month period of debate open to the entire population. At the end of the procedure, a report is drawn up for consideration by the body responsible for carrying out the project.[32] The *débat public* model inspired the Italian legislature to draft Article 22 of the Public Contracts Code for large-scale infrastructure projects,[33] according to which large-scale projects "with an impact on the environment, on towns and cities and on regional planning" are subject to the public debate procedure.[34]

The *États généraux*, on the other hand, consist of a series of consultations and public debates with broad participation, divided into various panels organised at the regional level, which precede the revision of the law on bioethics.[35] At the end of the work, the *Comité Consultatif National d'Ethique* has the task of drawing up a report summarising the results, which is sent to parliament for discus-

[30] See the report by the Loka Institute for Science & Technology of, by & for the people: http://loka.org/TrackingConsensus.html.

[31] The Policy paper *Democrazia partecipativa e legislazione regionale* by Eupolis Lombardia, published in June 2014 and available on the portal: www.polis.lombardia.it.

[32] See V. MOLASCHI, *La democratizzazione delle decisioni science and technology based,* cit., 478 ss. See also G. PEPE, *Il modello della democrazia partecipativa tra aspetti teorici e profili applicativi un'analisi comparata,* cit., 74 ff. and S. CASSESE, *La partecipazione dei privati alle decisioni pubbliche. Saggio di diritto comparato*, in *Rivista trimestrale di diritto pubblico*, 1, 2007, 19 ff.

[33] See the legislative decree no. 50, 18 April 2016, as amended by the legislative decree no. 56, 19 April 2017.

[34] On this issue, V. MOLASCHI, *Le arene deliberative, cit*., 258 ss.

[35] On the *États généraux* held in 2018, see L. GAFFURI, E. PULICE, Francia - Stati Generali di Bioetica: pubblicato il rapporto di sintesi del Comité consultatif national d'éthique, in www.biodiritto.org, 5 June 2018.

sion in the legislature.[36] This second paradigm appears to be particularly interesting ratione materiae: in fact, genetic screening and genomic medicine are among the bioethical issues that defined the scope of the 2018 *États généraux* debate.[37]

Within the heterogeneous practices mentioned above, some cross-cutting features can be identified that are particularly suited to deliberation on gene editing: first, these are places and procedures that allow for informing, as well as consulting and acquiring the opinion of, the majority of the individuals who are the recipients of a given decision or of as representative a sample of them as possible; second, "the effect of participation is not to transfer the final decision-making power to the participants",[38] nor to directly bind the legislature, but to open a public debate and allow the exchange of arguments that find their synthesis in a final product (be it a report, a budget or recommendations).

On the one hand, the fact that the outcome of the deliberation is not binding but at most carries out a "moral suasion capable of actually conditioning the formulation of public choices"[39] can be seen as an element of weakness of the participatory arenas. On the other hand, this characteristic helps to mitigate the objection that it would be inappropriate to entrust ordinary people (i.e. non-scientists) with decisions based on complex findings and knowledge.[40] Moreover, unlike a more incisive instrument, such as the popular referendum,[41] a possible rejection by the public of a certain technology does not close the matter in the long term. On the contrary, given the argumentative nature of deliberation, it helps to highlight the conditions under which it is possible to continue.

3. Genetic manipulation as a privileged field for public participation

In order to understand whether, and to what extent, public involvement can and should play an important role in decisions on gene editing regulation, it is necessary to take into account a number of factors that are peculiar to the technologies in question: the connection with environmental issues, the spatial and temporal extent of the effects that may arise from their application and, of course, the ethical sensitivity of the issues raised.[42]

This latter aspect, together with the divisiveness of the subject and the variety and relevance of the interests involved, has made the question of genetically modified organisms "the quintessential type of decision that the public has an explicitly stated desire to participate in".[43] This is a desire that is already understandable in relation to the genetic alteration of agricultural products – which has an im-

[36] Cfr. A. BLASIMME, *op. cit.,* 1067-1068.

[37] L. GAFFURI, E. PULICE, *op. cit*.

[38] R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa*, *cit*., 4.

[39] G. PEPE, *La democrazia partecipativa ambientale tra ordinamenti sovranazionali ed ordinamento italiano*, in *Federalismi.it*, 2, 2020, 183.

[40] "Should leaders listen […] to the public, some of whom may be convinced their last Whopper contained a Frankenfood patty because an Instagram influencer told them so?" (N. CURATO, S. NIEMEYER, *op. cit*., 2).

[41] One example is the 1987 referendum that effectively brought an end to the use of nuclear energy in Italy. A second referendum in 2011 ruled out the possibility of a new nuclear programme on our planet.

[42] This is especially true of their application to humans: suffice it to say that the Universal Declaration on the Human Genome and Human Rights, adopted on 11 November 1997 by the 29th session of the UNESCO General Conference, speaks of the human genome as the "heritage of humanity". (art. 1).

[43] T. ETTY, *Biotechnology*, in *The Yearbook of European Environmental Law*, 5, 2005, 314.

pact on food, the environment and the economy – and is destined to grow in relation to the use of techniques that allow intervention in animal and human DNA, given the greater impact they can have on people's lives and futures.

According to some, this demand for involvement should be protected as a real right under international law[44] – "the human right to science".[45] It should be understood not only as the right to benefit from scientific progress, but also as the right to "real participation in scientific life, which could also be understood as the right to be involved in the definition of research activities".[46]

Moving from the dimension of individual interest in participation to the broader horizon of benefit for the common good, we need to remember and treasure the lesson of Asilomar. As is well known, in the mid-1970s, Asilomar State Beach hosted a very important conference on the risks of recombinant DNA, which involved biologists, doctors and lawyers and led to the drafting of a series of guidelines on the "safe" use of genetic modification techniques and the production of GMOs. Although the initiative had the merit of demonstrating the sense of responsibility of the scientists involved, who participated with the aim of identifying the limits to which they voluntarily submitted when carrying out their activities, it has been severely criticised for excluding "laypeople" from the debate.[47] This prevented ethical and value-based arguments, which are necessary to establish "what forms of progress are culturally and morally acceptable",[48] from finding their rightful place alongside the assessment of scientifically measurable risks. Furthermore, it is legitimate to ask whether the deep divisions and contestations that still characterise the debate on GMOs could have been mitigated if the public had been properly informed and consulted from the outset.[49] It is precisely the lesson of Asilomar that has led authoritative doctrine to hope that future deliberations on gene editing (and CRISPR in particular) will be based on a rethinking of the relationship between science and democracy.[50] The same technological development could benefit from such a rethink if it resulted in less aversion on the part of the public, which – when not adequately informed – tends to view progress with fear.

A second element that makes gene technology an area of interest for deliberativism is its contact with the environment. The debate on environmental democracy[51] and participation in decision-

[44] See in particular Art. 27(1) of the Universal Declaration of 1948 ("Everyone has the right freely to participate in the cultural life of the community, to enjoy the arts and to share in scientific advancement and its benefits"), as well as Art. 15(1) of the International Covenant on Economic, Social and Cultural Rights of 1966 ("The States Parties to the present Covenant recognize the right of everyone: (a) To take part in cultural life; (b) To enjoy the benefits of scientific progress and its applications; (c) To benefit from the protection of the moral and material interests resulting from any scientific, literary or artistic production of which he is the author").

[45] M. MANCISIDOR, *Is there such a Thing as a human right to science in international law?*, in *ESIL Reflections*, 4, 2015.

[46] C. FLAMIGNI, *Sul consenso sociale informato*, in *Biolaw Journal*, 2, 2017, 202.

[47] S. JASANOFF, J.B. HURLBUT, K. SAHA, *CRISPR democracy: gene editing and the need for inclusive deliberation*, in *Issues in Science and Technology*, 32, 1, 2015.

[48] *Ibidem*, 3.

[49] Similarly, *ibidem*, 5.

[50] *Ibidem*, 3.

[51] See *ex multis* M. MASON, *Environmental Democracy: A Contextual Approach*, Londra, 1999; C. PITEA, *Diritto internazionale e democrazia ambientale*, Napoli, 2013; G. PAROLA, *Environmental Democracy at the Global Level:*

making processes concerning the environment has been ongoing for a long time and has led to important results, particularly at the level of international law:[52] we need look no further than the approval of the Aarhus Convention on access to information, citizen participation and access to justice in environmental matters,[53] implemented at the European level by EC Regulation 1367/2006. The forms taken by democratic participation in environmental decision-making do not always follow the patterns of participatory democracy but, thanks also to the Aarhus Convention,[54] the environment is undoubtedly considered "a context suited to welcoming the new frontiers of participation that deliberative democracy expresses".[55] In fact, this sector, "revealing impatience with traditional procedural participation, has, over the years, demanded forms of broader dialogue between institutions and members of the public with a view to acquiring and balancing the multitude of interests involved"[56] and, as we have seen, the deliberative arenas described in the previous paragraph are precisely in the direction of this dialogue.

The fact that the cultivation of GMOs falls within this context is beyond dispute, and in recent years, the European Union's Court of First Instance[57] has ruled that decisions relating to their commercialisation can also be considered matters of environmental law.[58] It follows that, within the EU, the so-called participation rights recognised by the Aarhus Convention and the Regulation also come into play in the process of authorising the marketing of GMOs. It is to be expected that the scope of these rights will be further extended in the future as gene technology progresses. To some extent, this has already happened, again through the courts, when in 2018 the Court of Justice of the European Union ruled that, as far as EU law is concerned, both products obtained by transgenesis (i.e. by inserting foreign DNA into the organism) and those obtained by mutagenesis are subject to the rules governing GMOs.[59] The Court also made it clear that the obligations relating to GMOs also apply to organ-

Rights and Duties for a new Citizenship, Londra, 2013; G. PEPE, *La democrazia partecipativa ambientale tra ordinamenti sovranazionali ed ordinamento italiano, cit.*, 179 ss.

[52] International law has also had the merit of emphasising the importance of public participation in the promotion of Sustainable Development (cf. para. 43 of UN Resolution A/RES/66/288 "The Future We Want"). This is also evident in the 2030 Agenda for Sustainable Development and, in particular, in Goal 16. See G. RAGONE, *The GMO Authorization Procedure in EU: Inclusivity, Access to Justice and Participation in Decision-Making, in Diritto Pubblico Europeo Rassegna online*, 2, 2019, 206 ss.

[53] See S.T. MCALLISTER, *The Convention on Access to Information, Public Participation in Decision-Making, and Access to Justice in Environmental Matters*, in *Colorado Journal of International Environmental Law and Policy*, 1999, 187 ss.; M. LEE, C. ABBOT, *The usual suspects? Public participation under the Aarhus Convention*, in *Modern Law Review*, 66, 2003, 80 ss.; M. PALLEMAERTS (a cura di), *The Aarhus Convention at Ten: interactions and tensions between conventional international law and EU environmental law*, in *Europa law publishing*, 2011.

[54] And in particular to its Second Pillar.

[55] V. MOLASCHI, *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico,* cit., 464.

[56] G. PEPE, *La democrazia partecipativa ambientale tra ordinamenti sovranazionali ed ordinamento italiano,* cit., 183.

[57] *TestBiotech et al. v. Commission* (T-177/13), 15 December 2016.

[58] see G. RAGONE, *Il delicato ruolo del giudice tra valutazioni scientifiche controverse e scelte politico-discrezionali in materia di OGM*, in *DPCE online*, 4, 2016, 252.

[59] *Confédération paysanne et al.* (C-528/16), 25 July 2018. See M.C. ERRIGO, *Diritto e OGM. Una storia complicata*, in *BioLaw Journal*, 1, 2020, 304.

isms obtained by means of mutagenesis techniques that emerged after the adoption of the reference legislation, as in the case of CRISPR.

If the environment is fertile ground for deliberativism, it is also because, in this context more than in others, it is necessary to "realise that [law] is no longer only the product of institutionalised popular sovereignty but is also the result of an increasingly non-institutionalised popular sovereignty".[60] In fact, while traditional representative institutions are responsible for decisions that affect the territories from which they derive legitimacy, choices concerning the environment generally have a broader scope, transcending territorial boundaries and affecting a population that does not necessarily belong to the same legal system.

The links between deliberativism and globalisation have been highlighted by various scholars. Some point out how the processes of globalisation, by contributing to the crisis of traditional representative institutions,[61] have the consequential effect of favouring the spread of deliberativist theories. Others, observing the shift from government to governance fostered by globalisation, suggest that – in the absence of representative institutions of global dimensions – deliberative arenas are the ideal place to discuss decisions that may have global reach.[62] This is the case in the regulation of impactful technologies, such as gene editing, including those applications that cannot be traced back to environmental matters and particularly those related to the human genome: "the human genome is not the property of any particular culture, nation or region; [...] It belongs equally to every member of our species".[63] This awareness, together with the imponderability of the magnitude of the effects of interventions on human DNA over time (especially when conducted in the germ line), has led to the assertion that "decisions about how far we should go in tinkering with it have to be accountable to humanity as a whole".[64]

But how can we consult citizens from all over the world and have a dialogue with them on the limits to which such complex technologies can be pushed? The Global Citizens' Assembly in Canberra is a first attempt to meet this challenge.

4. The case of the Global Citizens' Assembly on Genome Editing

The convening of a global citizens' assembly on genomic manipulation requires addressing problematic issues and implementation limitations that are not easy to overcome.[65] In particular, how should

[60] R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa, cit*., 9.

[61] See for instance G. PEPE, *Il modello della democrazia partecipativa tra aspetti teorici e profili applicativi un'analisi comparata,* cit., 13 ss.

[62] J.S. DRYZEK, A. BÄCHTIGER, K. MILEWICZ, *Toward a Deliberative Global Citizens' Assembly, cit.*, 33 ss.; J.S. DRYZEK et al., *Global citizen deliberation on genome editing, cit*., 1435 ss.

[63] S. JASANOFF, J.B. HURLBUT, K. SAHA*, op. cit.*, 2.

[64] *Ibidem*, 2.

[65] More generally, on the critical aspects of deliberative assemblies, see E. CAROLAN*, op. cit.*, passim. The Author, referring to the actual deliberation phase, emphasises the non-neutral nature of choices regarding "how to frame an issue; in what order it should be approached; from what disciplinary perspectives; using what kinds of 'evidence' and 'experts'; and subject to what limitations; shape how the process develops". All of these elements "inevitably privilege particular kinds of input and require difficult trade-offs between abstract delibera-

participants be selected? How should the work be organised and by whom should it be directed? What impact can be expected from such an experiment?

In general, the aspect of audience selection is one of the thorniest issues[66] since, in addition to having a significant impact on the success of the initiative, it represents a clear paradox: with participatory democracy, "one aspires to include everyone, but – in fact – one manages to actually involve only a few".[67]

The methods usually employed in deliberative arenas can be grouped into three archetypes, which can be applied in a mixed way: the open-door method, the microcosm and the mini-public.[68]

The open-door mechanism consists of self-selection: any citizen who so wishes and is informed of the existence of a particular arena can take part until a sufficient number of participants has been reached. The second method, the microcosm, presupposes that an organiser from outside the forum selects the participants, trying to create as faithful a representation as possible of all those who have a specific interest in the deliberation. In this case, the participants end up being the bearers of a pre-established vision and give rise to a 'hot' discussion.[69] This is an advantage from the point of view of the citizens' knowledge of the subject, but is detrimental to the possibility of the argumentative exchange being genuine and fruitful. Lastly, the archetype of the mini-public[70] involves the random selection of a sample of the population, possibly stratified in socio-demographic quotas (by gender, age, origin, etc.): "the bet is that any citizen, put in a position to talk to others and to acquire the necessary information, will be able to express precise positions on any public problem and build, together with others, intelligent solutions".[71] This type of selection leads to a debate that tends to be free of partisan interests[72] and is the most common in consensus conferences and citizens' assemblies. The Australian Global Citizens' Assembly is no exception.

In this case, the draw aims to select between 24 and 100 participants (depending on the scale of funding available), with people from all continents of the world, reflecting the global community as accurately as possible in terms of age, gender, education level, ethnicity and residence. If funding allows, the global assembly will be preceded by national forums, in which case some of the participants may be drawn from those taking part in the preparatory arenas. Once the panel of participants has been constituted, the work will be organised and led by the University of Canberra's Deliberative Democracy and Global Governance Centre, although the venue will not necessarily be in Australia. It is quite usual for this kind of conference to be followed step by step by deliberation professionals who can ensure that all stages of the work are carried out correctly.

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tive goods like expertise, independence, experience, impartiality, participation, openness and evidential scrutiny".

[66] See also L. PELLIZZONI, *op. cit.*, 150

[67] L. BOBBIO, G. POMATTO, *op. cit.*, 52.

[68] See R. BIFULCO, *Democrazia deliberativa e democrazia partecipativa*, *cit.*, 5; L. BOBBIO, G. POMATTO, *op. cit.*, 52-53.

[69] *Ibidem*, 58.

[70] Term first used in A. FUNG, *Recipes for public spheres: Eight institutional design choices and their consequences*, in *Journal of Political Philosophy*, 3, 2003, 338 ss.

[71] L. BOBBIO, G. POMATTO, *op. cit.*, 56-57.

[72] On the advantages and disadvantages of "cold" deliberations, see L. BOBBIO, G. POMATTO, *op. cit.*, 58.

In the given case, the programme foresees five working days. The first day will be devoted to the presentation of the rules of operation of the conference and to getting to know the participants. The next three days will be devoted to discussing the genetic modification of plants, animals and humans, respectively. The discussion will be conducted in groups in the presence of ethicists, legal experts and scientists, who will be asked to share their knowledge. On the fifth and final day, participants will be asked to draw up a document containing practical recommendations for decision makers, as well as for the scientific community, identifying the ethical issues they consider most problematic and the regulatory principles they consider most appropriate. The resulting synthesis document will be presented not only to national authorities and legislators but also to global players (such as the secretary-general of the United Nations and the directors-general of the WHO and FAO).

Although the final report has no legal force and will be amended at a later date to bring the debate up to date with the progress of genetic research,[73] it will provide decision-makers with a point of view that would be difficult to obtain in any other way: that of the citizens – the global community – who will be affected by the decisions.

5. Conclusions

The innovative reach of genetic engineering technologies, which can be applied to all forms of life on the planet (including humans), makes it necessary to reflect on which decision-making processes are best suited to influence what has been defined as "our genomic future".[74] Here, we have chosen to consider the opportunity to involve the public through participatory tools based on the principles of deliberative democracy, focusing in particular on the model offered by so-called consensus conferences. As we have seen, unlike the institutions of direct democracy, "these spaces of participation and deliberation [...] do not have, and cannot claim, any direct decision-making power, but this does not make them irrelevant".[75]

Among the merits that have been recognised in these kinds of democratic experiences is that they open up a dialogue between institutions and civil society.[76] This can "play a decisive role in legitimising (or critically, de-legitimising) the decisions taken by the institutions",[77] enriching the democratic nature of our legal systems.[78] Dialogue with the public in the phase preceding the actual decision-making moment can also contribute to the undertaking of better choices: decision-makers can, in fact, base their decisions on considerations other than those offered to them by experts in the field.

[73] J.S. Dryzek et al., *Global citizen deliberation on genome editing, cit*., 1437.
[74] A. Greenfield, *Our genomic future*, in *News Scientist*, 241, 2019, 24-25.
[75] A. Floridia, *Democrazia partecipativa e democrazia deliberativa: una risposta plausibile alla "crisi della democrazia"?, cit*., 3.
[76] J.S. Dryzek et al., *Global citizen deliberation on genome editing, cit*., 1436.
[77] *Ibidem*, 3.
[78] R. Bifulco, *Democrazia deliberativa e democrazia partecipativa*, *cit*., 7.

In short, if the decisions concerning these technologies were anticipated by a public discussion, they would enjoy greater legitimacy,[79] and there would be a way to bring out – and, in time, correct – the criticalities perceived by the public. Of course, as has already been pointed out, the outcome of the deliberation will not necessarily correspond to an improved impact on the final decision; this is due both to the possible inexperience of the participants and to the non-binding nature of their recommendations. In a certain sense, the relationship between the citizens participating in the deliberation and the final decision makers is comparable to that between the amicus curiae and the courts: their opinion may be ignored but, at the same time, their point of view enters the cognitive horizon of the subject who has the right to the last word.

A further beneficial effect of the establishment of deliberative arenas for emerging scientific and technical issues is the reduction of anti-scientific populism by increasing public confidence in how policy-makers use expert advice.[80] As seen in the management of the global emergency following the COVID-19 pandemic, on the one hand, the role of experts (and committees with the appropriate scientific and technical expertise) is crucial to support decision-making in certain areas; on the other hand, the internal divisions within the scientific community, the opacity of the criteria for relying on a certain vision, and the instability of scientific findings, which are subject to continuous revision and re-evaluation over time, may cause scepticism among the public about the validity of the decisions made.

Allowing a section of the population affected by a given innovation to ask questions and talk to scientists and experts, besides being an interesting example of co-production between science and society,[81] helps to create an "informed social consensus".[82] This principle, which has been "knocking at the door of bioethics for some time"[83] and which expresses an ideal relationship between citizens and scientific research, "can play a role in guaranteeing and promoting science at a time when its daily progress is giving rise to growing hopes and concerns".[84]

In conclusion, it seems possible to say that the potential of the creation of places of public participation in science and technology-based decisions, such as the Global Citizens' Assembly announced by the Centre for Deliberative Democracy and Global Governance, is to take steps towards "overcoming the crisis of legitimacy from which scientific and political institutions suffer".[85] This suggests the need to continue the debate on deliberativism and its concrete ways of implementation in order to contribute to overcoming its limitations, which we have tried to highlight over the course of the discussion.

[79] The stronger legitimacy of decisions preceded by public consultation makes it necessary to reflect, secondly, on the modalities and the degree of pervasiveness that judicial review can adopt on them. Regarding the models of control by the courts on participation procedures, see S. CASSESE, *op. cit*., 18 ss.

[80] See V. MOLASCHI, *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico, cit.*, 469 and A. BLASIMME, *op. cit*., 1068.

[81] Cfr. M. TALLACCHINI, *op. cit*., passim.

[82] On this notion, see C. FLAMIGNI, *op. cit*., passim.

[83] *Ibidem*, 202.

[84] *Ibidem*, 201.

[85] V. MOLASCHI, *La democratizzazione delle decisioni science and technology based. Riflessioni sul dibattito pubblico, cit*., 470.

Special issue

Children's civil liability actions regarding parental germinal gene-editing decisions

Ekain Payán Ellacuria*

ABSTRACT: Advances in reprogenetic tools have completely changed parents' previously passive role in deciding what characteristics their children will have, giving rise to significant bioethical and legal implications. They now have genetic information which they can use in their reproductive decision-making to avoid serious disease in their children. Thus, children might bring claims against their parents, not only for their actions but also for their omissions. Therefore, this paper discusses whether parental freedom jeopardizes the rights of children and future generations. It discusses various conflicting and introduces the current legal discourse around the feasibility of CRISPR-Cas9 lawsuits.

KEYWORDS: Germline gene-editing, non-directive counselling, procreative autonomy, procreative beneficence, serious disease

SUMMARY: 1. Introduction – 2. Prospective parents' reproductive rights and responsibilities – 3. Civil liability actions: Wrongful birth and wrongful life – 4. The legal issue: Claims of prenatal damage for germline gene-editing – 5. Final remarks.

1. Introduction

Throughout human history, humans have desired to acquire new capabilities[1] but often lacked the ability to do so. In ancient times, it was believed that newborns received particular characteristics because of God's will, so parents had no choice but to submit to what was a genetic health lottery[2] and accept their children as they were. However, assisted reproductive technologies (ART) have opened up the possibility of artificial reproduction, which grants couples new rights and freedoms. Thus, if one of partner is unable to procreate, such as someone suffering from infertility, it is possible to use donated sperm or eggs, surrogate gestation, adopt, and conduct preimplantation genetic diagnosis (PGD) or prenatal diagnosis (PD) and select the desired embryos, allowing people to select for healthier biological offspring. However, gene therapy is the only method for correcting rare and incurable inherited disorders in both the unborn and

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[1] N. BOSTROM, *A history of transhumanism thought*, in *Journal of Evolution and Technology*, 14, 1, April 2005, 1.

[2] M. SANDEL, *The case against perfection,* Boston, 2004, 145.

subsequent generations.[3] Consequently, it seems inevitable that gene-editing will be used in the near future.[4]

A milestone in human reproduction occurred in late 2018 with the birth of the first two genetically edited human beings.[5] The purpose of this editing was not to cure an illness but rather to increase their resistance to acquiring human immunodeficiency virus (HIV) because their father was HIV-positive. This change can cause unforeseen and harmful lifelong consequences for children, such as off-target effects or mosaicism. He Jiankiu announced that he will monitor the children throughout their lives. However, the editing of their genes may have violated their fundamental rights, so the parents may bring a claim against the doctor for failing to secure properly informed consent[6] and advising them to receive genetic counselling before they agreed to the editing. When they reach the appropriate age, the children themselves may sue healthcare personnel if their parents have not done so on their behalf already or may even sue their own parents for causing them injury by their choice to have their genes edited.

The latter situation has already occurred for other families and, although the literature has addressed the bioethical implications of gene-editing,[7] it has not sufficiently examined compensatory liability for damages to human embryos. The goal of this paper is to identify parents' reproductive rights and responsibilities and what legal actions children can take against their parents and healthcare personnel for gene-editing under the current legal framework and precedents. It also addresses a double dilemma by focusing on the rights and guiding principles for how to treat children.

2. Prospective parents' reproductive rights and responsibilities

The right to procreate is not established per se by law. Nevertheless, it can be inferred from some international treaties that are binding on the signatory states. For example, the *Convention for the Protection of Human Rights and Fundamental Freedoms* (1950) Art. 3 prohibits inhuman and degrading treatment, Art. 8.1 recognizes the right to respect family life, and Art. 12 grants the right to start a family. The *International Covenant on Civil and Political Rights* (1966) Art. 23.2 grants the right to start a family. Therefore, the right to start a family implicitly protects procreative and childrearing rights. Thus, the freedom to decide whether to have children or not[8] can only be limited

[3] R. ANDORNO, A.E. YAMIN, *The right to design babies? Human rights and bioethics,* in *OpenGlobalRights*, January 2019, accessible on https://www.openglobalrights.org/the-right-to-design-babies-human-rights-and-bioethics/, (last visited 15/04/2021).

[4] J.A. DOUDNA, *The promise and challenge of therapeutic genome editing,* in *Nature,* 578, February 2020, 229.

[5] M. MARCHIONE, *Chinese researcher claims first gene-edited babies,* in *Associated Press,* November 2018, accessible on https://apnews.com/4997bb7aa36c45449b488e19ac83e86d, (last visited 15/04/2021).

[6] P. SANTILLÁN-DOHERTI, P. GRETHER-GONZÁLEZ, M.J. MEDINA-ARELLANO, S. CHAN, R. TAPIA-IBARGÜENGOITIA, I. BRENA-SESMA, et. al., *Considerations on genetic engineering: regarding the birth of twins subjected to gene edition,* in *Gaceta Médica de México*, 156, 2020, 54-55.

[7] D. ARCHARD, P. DABROCK, J.-F. DELFRAISSY, *Human-genome editing: ethics councils call to governments worldwide,* in *Nature*, 579, March 2020, 29.

[8] J.A. ROBERTSON, *Procreative Liberty and the Control of Conception. Pregnancy, and Childbirth*, in *Virginia Law Review*, 69, 3, 1983, 406.

by public authorities in justified and exceptional circumstances, which include when such freedom would harm third-party interests.[9] In addition, reproductive freedom is linked to the guarantees of individual autonomy and free development of the personality by the 1948 *Universal Declaration of Human Rights* Arts. 26.2 and 29.1 (UDHR), personal identity by the 1997 *European Convention on Human* Rights and Biomedicine Art. 1 (ECHRB), human dignity by the UDHR Art. 1 and the 2000 *Charter of Fundamental Rights of the European Union*, and well-being,[10] all of which have inspired several democratic constitutions. The volume and importance given to related guarantees are the reasons why reproductive liberty is commonly considered a subjective right.[11]

However, this freedom is not absolute. For example, parents have a duty to take care of their children and provide them with food, education, and healthcare, duties which remain throughout their minority and beyond if their children are unable to provide this for themselves because they are disabled when they reach the age of majority. Arts. 1 and 6 of the 1997 Educational, Scientific and Cultural Organization's (UNESCO) *Declaration on the Responsibilities of the Present Generations Towards Future Generations* hold that people have responsibilities towards future generations, so they must respect the human genome as part of respecting human dignity and human rights. This legal good was further enshrined in the 2005 UNESCO *Universal Declaration on Bioethics and Human Rights* Art. 16 which addresses the impact of life sciences on future generations. However, despite the other articles mentioned above, some argue that Art. 16 this is a staging of the sanctification of the heritage of humankind because germline gene-editing (GGE) is constantly changing and so would not introduce any novelty into the gene pool.[12] Furthermore, ECHRB Art. 2 gives puts the interests and welfare of individuals over those of society and science. Thus, according to some interpretations, the responsibility towards future generations[13] and the requirement to protect human dignity[14] would require that action be taken to change children's genes in a way that would make them healthier.

Be that as it may, parents make decisions for their children before they are born and until they reach the age of majority during which time the children are deemed to be incapable of giving informed consent to medical procedures,[15] giving parents both rights and obligations.

[9] G. Cavaliere, *The problem with reproductive freedom. Procreation beyond procreators' interests,* in *Medicine, Health Care and Philosophy*, 23, 2020, 132.

[10] J.A. Robertson, *Children of choice: Freedom and the new reproductive technologies,* Princeton, 1994, 24.

[11] I. Alkorta Idiakez, *Nuevos límites del derecho a procrear,* in *Derecho privado y Constitución*, 20, 2006, 13.

[12] A.L.V. Hammerstein, M. Eggel, N. Biller-Andorno, *Is selecting better than modifying? An investigation of arguments against germline gene-editing as compared to preimplantation genetic diagnosis,* in *BMC Medical Ethics,* 20, 83, 2019, 4.

[13] I. de Miguel Beriain, J. Almqvist, *Ethical questions in gene therapy,* in J. Faintuch, S. Faintuch (eds.), *Precision Medicine for Investigators, Practitioners and Providers,* San Diego, 2020, 526.

[14] I. de Miguel Beriain, B. Sanz, *Human dignity and gene-editing: Additional support for Raposo's arguments,* in *Bioethical Inquiry,* 17, 2, 2020, 167.

[15] T. Ishii, I. de Miguel Beriain, *Safety of germline genome editing for genetically-related "future" children as perceived by parents,* in *The CRISPR Journal*, 2, 6, 2019, 371.

3. Civil liability actions: Wrongful birth and wrongful life

The obligation to compensate victims of medical malfeasance may arise for contractual reasons or from non-contractual reasons, such as negligence. However, both types of reasons must involve the performance of an unlawful act, omission, fault, or negligence that caused damage.[16] Private law claims can be supported by several allegations, such as wrongful conception, wrongful birth, wrongful life, and wrongful death, but birth and life actions will be briefly explained below because of their repeated application and relation to factual assumptions.

Both parents jointly or the mother solely can bring a claim for wrongful birth against obstetricians who do not propose appropriate diagnostic tests or detect foetal aberrations, depriving them of the opportunity to legally terminate their pregnancy. The damages of such claims may include the costs of raising the child, costs of providing extra care that certain children might require, and non-material losses.[17] Furthermore, the child or their representatives, such as their parents or legal guardians, can bring wrongful life claims against physicians who allowed them to be born to a life of suffering that caused them to have preferred to have not been born.

The main differences between wrongful birth and wrongful life claims are who brings them and their legal bases, even though parents often bring both of them.[18] Another difference is that wrongful birth claims are generally admitted while wrongful life claims are generally denied.[19] The main reason that the rights to health and human dignity are considered fundamental in most Western countries is because of the assumed legal, ethical, and social obligations to protect life. The assumption is that the lives of those suffering from pathologies are no less valuable than those that are not. Another reason that they refuse is that not doing so could lead to defensive medicine.[20] Regardless of the reason, the strength of this assumption differs by country and there are a few jurisdictions that do recognize a child's difficulties in life as grounds for bringing a legal claim (e.g., the Netherlands: Dutch Supreme Court, 18 March 2005, *Kelly case*;[21] California, USA: *Curlender v. Bio-Science Laboratories* (1980),[22] *Turpin v. Sortini* (1982);[23] Washington, USA: *Haberson v. Parke-Davis* (1983);[24] New Jersey,

[16] A. Emaldi Cirión, *El consejo genético y sus implicaciones jurídicas*, Bilbao-Granada, 2001, 261-264.

[17] A. Vicandi Martínez, *El concepto de wrongful birth y su inherente problemática. Una polémica del pasado y del presente*, in *Revista de Derecho, Empresa y Sociedad (REDS)*, 3, 2013, 50-54.

[18] A. Emaldi Cirión, *op. cit*., 239.

[19] A. Macía Morillo, *Una visión general de las acciones de responsabilidad por "wrongful birth" y "wrongful life" y de su tratamiento en nuestro ordenamiento jurídico*, in *Anuario de la Facultad de Derecho de la Universidad Autónoma de Madrid (AFDUAM),* 10, 2006, 90.

[20] I. Giesen, *Of wrongful birth, wrongful life, comparative law and the politics of tort law systems*, *Tydskrif vir Heedendaagse Romeins-Hollandse* Reg, 72, 2009, 267; C. M. Romeo Casabona, A. Perin (eds.), *Derecho y medicina defensiva: legitimidad y límites de la intervención penal,* Bilbao-Granada, 2020, 1-24.

[21] HR 18 March 2005, 2006 Nederlandse Jurisprudentie 606 nt JBMV (Kelly).

[22] *Curlender v. Bio-Science Laboratories*, 106 Cal. App. 3d 811,165 Cal. Rptr. 477, hearing denied, No. 2 Civ. 58192 Div. 1 (Cal. September 4, 1980).

[23] *Turpin v. Sortini*, 119 Cal. App. 3d 690,174 Cal. Rptr. 128, 1981, rev'd, May 3, 1982, No. S.F. 24319.

[24] *Haberson v. Parke-Davis*, 98 Wash. 2d 460, 656 P.2d 483, January 6, 1983.

ISSN 2284-4503

USA: *Prokanic v. Cillo* (1984))[25]. Many jurisdictions have unclear legislation, including Belgium, Japan, and Spain,[26] although in Spain, wrongful life is contrary to the legal order.[27]

Given this legal background, children with congenital diseases may sue not only the medical personnel involved in their being born but also their parents who decide to continue with their birth knowing that they have a congenital disease or who reject embryonic treatment that could cure congenital diseases. In this context, the next section will discuss how GGE may change the concept of parental responsibility as it has been understood so far, leading to the occurrence of situations unanticipated by the law.

4. The legal issue: Claims of prenatal damage for germline gene-editing

At first glance, GGE would seem to be prohibited. The *Charter of Fundamental Rights of the European Union* Art. 3.2 bans eugenic practices, so given that GGE edits genes, some could see it as a slippery slope to eugenics.[28] However, some argue that what separates it from eugenics, which was characterized by bad science, state intervention, and discrimination, is knowledge, a lack of coercion, and a therapeutic intent.[29]

Furthermore, Art. 13 of the ECHRB requires that parents not intend to modify the genes of their offspring, *Directive 98/44/EC* on the legal protection of biotechnological inventions Art. 6.2.b prohibits the alteration of germline genetic identities, and *EU Regulation No. 536/2014* on clinical trials on medicinal products for human use Art. 90. However, these statutes mention genomes and germlines, which are different, so they may permit GGE.[30]

Furthermore, a global ban on GGE could infringe children's fundamental right to health in terms of equitable access to health benefits guaranteed by ECHRB Art. 3, the right to benefit from biological and medical progress guaranteed by UDHR Art. 27.1 and UNESCO's *Universal Declaration on the Human Genome and Human Rights* (UDHGHR) Art. 12.a, and the freedom of research guaranteed by UNESCO's UDHGHR Art. 12.b and ECHRB Art. 15, among others.

Another reason that gene-editing is feasible is that there are rules that expressly establish the individual right to claim compensation for undue damage. In this regard, UNESCO's UDHGHR Art. 8 states that "Every individual shall have the right, according to international and national law, to just reparation for any damage sustained as a direct and determining result of an intervention affecting

[25] *Prokanic v. Cillo*, 97 N.J. 339, 478 A.2d 755, August 1, 1984.

[26] P. Frati, V. Fineschi, M. Di Sanzo, R. La Russa, M. Scopetti, F. M. Severi, et. al., *Preimplantation and prenatal diagnosis, wrongful birth and wrongful life: A global view of bioethical and legal controversies,* in *Human Reproduction* Update, 23, 3, 2017, 346-348.

[27] J. Mª Fugardo Estivill, *Procreación humana y acciones de responsabilidad*, Barcelona, 2018, 158.

[28] I. Brown, *The new eugenics and human progress,* in *Journal of Policy and Practice in Intellectual Disabilities*, 16, 2, 2019, 137-140.

[29] G. Cavaliere, *Looking into the shadow: The eugenics argument in debates on reproductive technologies and practices*, in *Monash Bioethics Review*, 36, 1-4, 2018, 14.

[30] I. de Miguel Beriain, E. Armaza, A. Duardo Sánchez, *Human germline editing is not prohibited by the Oviedo Convention: An argument*, in *Medical Law International*, 19, 2-3, 2019, 227-228.

his or her genome".[31] Likewise, ECHRB Art. 24 states "The person who has suffered undue damage resulting from an intervention is entitled to fair compensation according to the conditions and procedures prescribed by law".[32]

Given that the ban on GGE may not be absolute and there are statutes in place providing for financial compensation for the damage caused by its application, the next step is to determine whether children can bring actions for wrongful life against their parents and healthcare providers. As a preliminary premise for making this determination, it must be assumed that parents are not obliged to know the risk of transmitting a hereditary condition to their children, so they should not be held liable for negligence if they unknowingly do so. Even if they are aware of such risk, they should not be forced to genetically modify their children as they may not want to do so for moral or religious reasons. Compelling them to do so would violate their fundamental right to religious freedom as guaranteed by the UDHR Art. 18 and would make such genetic treatment more similar to eugenics. An economic argument can also be made for not compelling people to edit their children's genes. There will be people who may want to edit their children's genes, but will not be able to afford to do so, despite how relatively inexpensive clustered regularly interspaced short palindromic repeats (CRISPR-Cas9) procedures will likely become. To try to compel them to do so could constitute discrimination under UDHR Arts. 2 and 7 and ECHRB Art. 11 unless the cost of such procedures was borne by the national health system, something that is impossible in countries without such systems.

However, it is a different matter if both parents freely and voluntarily decide to use GGE. Assuming that GGE is effective and safe, children and even grandchildren might be able to bring civil claims against their families. One is positive and would be brought by children whose genes were edited against their will. The other is negative and would be brought when parents resorted to ART but not GGE.

The first type of claim calls into question the identity of the unborn and brings up the non-identity problem,[33] which entails the risk of giving birth to different individuals. However, some disagree because the embryo already has an identity by the time its genes are modified,[34] so illnesses, like Alzheimer's, are a part of that identity. Another argument against editing the genes of embryos is that it violates the child's right to autonomy as an adult.[35] Be that as it may, it seems ethically permissible for parents to use GGE to prevent serious diseases in their children, especially monogenic

[31] United Nations Educational, Scientific and Cultural Organization, *Universal Declaration on the Human Genome and Human Rights. Adopted by the General Conference of the United Nations Educational, Scientific and Cultural Organization at its twenty-ninth session on 11 November 1997; endorsed by General Assembly resolution 53/152 of 9 December 1998*, November 1997, accessible on https://www.ohchr.org/EN/ProfessionalInterest/Pages/HumanGenomeAndHumanRights.aspx, (last visited 15/04/2021).

[32] Council of Europe, *Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine*, *April 1997*, accessible on https://www.coe.int/en/web/conventions/full-list/-/conventions/treaty/164, (last visited 15/04/2021).

[33] D. PARFIT, *Reasons and Persons*, Oxford, 1987, 371-372.

[34] A. OMERBASIC, *Genome Editing, Non-Identity and the Notion of Harm*, in M. BRAUN, H. SCHICKL, P. DABROCK (eds.), *Between Moral Hazard and Legal Uncertainty. Ethical, Legal and Societal Challenges of Human Genome Editing*, Wiesbaden, 2018, 73-74.

[35] J. FEINBERG, *Freedom and Fulfilment: Philosophical Essays,* New Jersey, 1992, 76-97.

ones, such as Tay-Sachs, Huntington's, or cystic fibrosis.[36] This type of use has legal endorsement in the 1989 *Convention on the Rights of the Child* Art. 3, which stipulates that the best interests of the child must be pursued, and Art. 24, which stipulates that children have the right to the highest attainable standard of health both before and after birth. The latter right is also safeguarded by the 1966 *International Covenant on Economic, Social and Cultural Rights* Art. 12. Both of these rights are supported by the principle of procreative beneficence, which holds that parents have a moral obligation to make choices such that their children will have the best possible life, given that there is a considerable consensus on how illnesses affect expected well-being.[37] However, some hold that quality of life is a product of physical health as well as relationships, social inclusion, material well-being, and being able to exercise rights.[38] The sum of these elements in a person's life are what produces overall higher levels of happiness[39] and what ensures the diversity and equality of all humans.

In light of all of these facts and arguments, the children whose genes were edited may have reasonable grounds to sue their parents. Although their parents had their genes edited to reduce the chances that they would get HIV, HIV infection is no longer lethal and children had not yet actually been infected by it. Therefore, they were unnecessarily exposed to risk despite having safer alternatives, such as treatment with antiretroviral therapies, condom use, or sperm washing. However, the admissibility of this claim will depend on whether the parents gave informed consent to the procedure and received genetic counselling. If they did not, their consent would be invalid, so the doctor and his team would be liable to the children.

A second situation would be if the parents used an ART, such as PGD, instead of GGE and *in vitro* fertilization (IVF).[40] Even though some commentators have argued that both techniques achieve the same results, they are not equivalent. PGD examines embryos for genetic mutations and only healthy ones are transferred to the uterus. Defective embryos are discarded or used for research purposes. Scientists often defend PGD over other practices because of its safety, although it may not be fully effective when both parents have the same recessive monogenic disorder.[41] However, some people may highly value the embryos for ideological or religious reasons and[42] so refuse to deliberately lose

[36] GERMAN ETHICS COUNCIL, *Intervening in the Human Germline*, May 2019, 26, accessible on https://www.ethikrat.org/fileadmin/Publikationen/Stellungnahmen/englisch/opinion-intervening-in-the-human-germline-summary.pdf, (last visited 15/04/2021); NATIONAL ACADEMY OF MEDICINE, NATIONAL ACADEMY OF SCIENCES, THE ROYAL SOCIETY, *Heritable Human Genome Editing*, Washington, D. C., September 2020, 96-104.

[37] J. SAVULESCU, G. KAHANE, *The Moral obligation to create children with the best chance of the best life*, in *Bioethics*, 23, 5, June 2009, 279.

[38] J. REINDERS, T. STAINTON, T.R. PARMENTER, *The quiet progress of the new eugenics. Ending the lives of persons with intellectual and developmental disabilities for reasons of presumed poor quality of life,* in *Journal of Policy and Practice in Intellectual Disabilities*, 16, 2, 2019, 101.

[39] I. BROWN, R.I. BROWN, A. SCHIPPERS, *A quality of life perspective on the new eugenics,* in *Journal of Policy and Practice in Intellectual Disabilities,* 16, 2, 2019, 123.

[40] M. VIOTTI, A. R. VICTOR, D. K. GRIFFIN, J. S. GROOB, A. J. BRAKE, C. G. ZOUYES, et. al., *Estimating demand for germline genome editing: An in vitro fertilization clinic perspective,* in *The CRISPR Journal,* 2, 5, 2019, 304.

[41] R. RANISCH, *Germline genome editing versus preimplantation genetic diagnosis: Is there a case in favour of germline interventions?,* in *Bioethics,* 34, 1, 2020, 63.

[42] M. WALKER, *Eugenic selection benefits embryos,* in *Bioethics,* 28, 5, 2014, 215.

them.[43] Also, PGD after IVF requires mothers to undergo ovarian stimulation and invasive punctures to introduce the embryos into their wombs.[44] This process is more stressful and painful than the process that men undergo that requires them to abstain from sex for three days and provide a sperm donation through a mostly quick and painless process. Thus, PGD after IVF further burdens mothers in ways that it does not burden fathers. On the other hand, GGE can accurately and easily give health to a person who will one day exist. Also, GGE has no reproductive effects, unlike somatic gene-editing, which is applied to a patient's own cells, and prevents future diseases from arising, unlike PGD.[45] Moreover, over time researchers are likely to develop more control over and knowledge about CRISPR-Cas9 by improving techniques, such as prime editing[46] and chimeric antigen receptor T-cell therapies.[47]

In this case, the child would have the right to an open future but their best interests in terms of health would not be served and their right to enjoy the highest attainable standard of health would be violated. As mentioned above, human dignity and the rights of future generations can be used as arguments both for and against gene-editing, so the way that they are used depends on each individual and how they view the issue.

5. Final remarks

This paper discussed one of the edges of GGE. At present, this debate is currently moot given the state of technological development, but if GGE becomes safe and not subject to legal issues,[48] many more people would likely use it to treat their unborn children's diseases because it is easier and more effective to do so early in a person's life. However, GGE recipients may sue their parents for having edited their genes. Parents may mount the defences that such editing was therapeutic, its benefits outweighed its risks, and, under most legal regimes, parents are not liable for medical procedures which they consented for their children to undergo.[49] A good policy reason for exempting parents from liability for editing their children's genes is that they may opt to not reveal such editing if it would expose them to litigation.[50]

[43] A. Nordberg, T. Minssen, O. Feeney, I. de Miguel Beriain, L. Galvagni, K. Wartiovaara, *Regulating germline editing in assisted reproductive technology: An EU cross-disciplinary perspective*, in *Bioethics*, 34, 1, 2020, 18.

[44] F. Simonstein, *Gene-editing, enhancing and women's role,* in *Science and Engineering Ethics,* 25, 4, 2019, 1011.

[45] N. Kofler, K. L. Kraschel, *Treatment of heritable diseases using CRISPR: Hopes, fears, and reality*, in *Seminars in Perinatology*, 42, 8, 2018, 515-516.

[46] A.V. Anzalone, P.B. Randolph, J.R. Davis, A.A. Sousa, L. W. Koblan, J. M. Levy, et. al., *Search-and-replace genome editing without double-strand breaks or donor DNA,* in *Nature,* 576, 2019, 149-157.

[47] J.R. Hamilton, J.A. Doudna, *Knocking out barriers to engineered cell activity,* in *Science,* 367, 6481, 2020, 976-977.

[48] I. de Miguel Beriain, C.M. Romeo Casabona, *The regulation of human germline genome modification in Spain,* in A. Boggio, C.P.R. Romano, J. Almqvist (eds.), *Human Germline Genome Modification and the Right to Science. A Comparative Study of National Law and Policies,* Cambridge, 2020, 379.

[49] D. Krekora-Zając, *Civil liability for damages related to germline and embryo editing against the legal admissibility of gene-editing,* in *Palgrave Communications,* 6, 30, 2020, 6.

[50] T. Ishii, I. de Miguel Beriain, *op. cit.,* 374.

From the opposite perspective, children can take legal action against their parents for not editing their genes to help them avoid certain diseases. Nevertheless, some claim that no one has the right to be born healthy even if they could have been.[51] However, these people may change their minds as gene-editing technology develops and gene-editing is done to protect children's rights to life and good health.

For that to ever happen, all civil liability must disappear because it remains the main obstacle preventing gene-editing from becoming more common,[52] particularly with regard to the causal relationship between the transmission of a disease and the damage it causes.[53] The latter excludes situations in which someone may be disadvantaged, but not sufficiently so to create civil liability.

Opening the door even slightly to GGE may lead to an increase in litigation by children against their parents and healthcare providers, a scenario in which this contribution tries to deepen by setting out the various fatherly and filial rights and duties involved in order to provide guidance and legal certainty for lawyers.

[51] B.M. KNOPPERS, E. KLEIDERMAN, *Heritable genome editing: Who speaks for "future" children?*, in *The CRISPR Journal*, 2, 5, 2019, 289.

[52] A. EMALDI CIRIÓN, *op. cit.*, 261-264.

[53] J. Mª FUGARDO ESTIVILL, *op. cit.*, 216.

ISSN 2284-4503

When parents look for a "better" child (reproductive choices and genetic planning)

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ABSTRACT: The focus of this paper is on the selection of apparently neutral characteristics, refused in several jurisdictions, namely in Europe. The reasons for such refusals are considered in this paper. However, arguments such as the difficulty in distinguishing apparently neutral features from others that are health-related, the recognition that some features are not so neutral after all, and the discredit of genetic determinism are counterarguments that may lead to a different assessment of non-health related reproductive decisions. The premise of this paper is that the prohibition of parental reproductive choices for selecting non-health related features should be reassessed due to the fragility of the arguments invoked against them.

KEYWORDS: Enhancement; genetic determinism; offspring selection; reproductive decisions; reproductive rights

SUMMARY: 1. Introduction – 2. Parental reproductive choices in light of the existing regulations – 3. Classic arguments against NHRRD – 3.1. Eugenics – 3.2. Loss of genetic diversity – 3.3. Genetic discrimination – 3.4. Objectification of children – 3.5. Violation of the child's right to an open future – 3.6. The artificial over the natural – 4. Parental reproductive choices in light of reproductive rights – 5. Problems faced by this position – 5.1. The distinction between health-related features and other features – 5.2. The relevance of the selected features – 6. The admissibility of some NHRRD – 6.1. Reasons leading to the admissibility of NHRRD – 6.2. Features to be selected – 6.3. Offspring selection and the destruction of human life – 7. Final considerations.

1. Introduction

If science continues to develop, it is possible that reproductive techniques and genetic technologies will provide parents the possibility to select almost all genetic characteristics of their children. The paper will use the concept of "genetic selection" to refer to all medical and scientific procedures that allow to determine offspring features. The concept includes preimplantation genetic diagnosis (PGD), but also genetic modification (gene editing), which is also a form of selection, since by modifying the genetic code of their embryo/foetus parents can select the traits of the future child.[1] That same aim can be achieved – eventually less successfully - using other procedures, such as

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[1] Recognizing that despite the difference between genetic selection (the authors are referring to preimplantation genetic diagnosis) and genetic modification both proceedings have the same aim, A.L. v. HAMMERSTEIN, A.L., M. EGGEL, N. BILLER-ANDORNO, *Is Selecting Better Than Modifying? An Investigation of Arguments Against Germline Gene Editing as Compared to Preimplantation Genetic Diagnosis,* in *BMC Med Ethics*, 20, 2019.

ISSN 2284-4503

the selection of gamete donors[2] or abortion,[3] which will also be considered under the concept of "genetic selection".

The range of choices might cover not only features with direct therapeutic implications, but also non health-related features, such as the QI, and even apparently neutral features, such as the eye colour. Currently most of these parental choices are not scientifically possible, but eventually they will become possible in the (near?) future, imposing the debate on these matters. Curiously, some forms of genetic selection that might seem too futuristic are in fact about to become reality. For instance, the scientific community already knows that when the CCR5 gene is deactivated (as it happened in the infamous experiment carried out by He Jiankui with the Chinese twins Lulu and Nana)[4] it can lead to increase learning ability.[5]

This paper will only deal with parents' choices to select features of their children that do not relate – at least, not directly – with health, hereafter defined as Non-Health Related Reproductive Decisions (NHRRD). Mainstream scholars[6] argue that parents should be allowed to select features related with the health of their progeny (for instance, abort a foetus with a severe malformation or select a certain gender for their child to avoid a disease associated to the opposite gender), but not to select other kind of features. Lawmakers around the world tend to follow this solution. The paper will focus its analysis on European countries, but references to other jurisdictions will also be made (part 2).

The paper will start by analysing the arguments commonly invoked against NHRRD, namely eugenics, the loss of genetic diversity, genetic discrimination, the commodification of the child in a way that he/she becomes an object of the parent's wishes, limitations on the child's free development (right to an open future), and the supremacy of elements coined by nature over man-made elements. As the paper will demonstrate, none of these arguments have real substance (part 3).

Encouraged by the failure of classic arguments, some authors (the position of John Robertson is paradigmatic in this regard)[7] have suggested that NHRRD are not undermined by these arguments,

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[2] For instance, see M.J. McGinniss, M.A. McGinniss, *Carrier Screening and Heterozygote Testing*, in R.E. Pyeritz, B.R. Korf, W.W. Grody (eds.), *Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics*, 7th edition, San Diego, 2018, 283-298, describing gamete donation as a form of selection against certain diseases.

[3] Referring the similarity of aims between abortion and PGD see C. Cameron, R. Williamson, Is *There an Ethical Difference Between Preimplantation Genetic Diagnosis and Abortion?,* in *J Med Ethics*, 29, 2, 2003, 90-92.

[4] About this incident see V.L. Raposo, The First Chinese Edited Babies: A Leap of Faith in Science, in *JBRA Assisted Reproduction*, 23, 2019, 3 197-199.

[5] There is a wide range of literature about this. See, for instance, D. Cyranoski, *Baby Gene Edits Could Affect a Range of Traits*, in *Nature*, 2018; M.T. Joy, E.B. Assayag, D. Shabashov-Stone et al., *CCR5 Is a Therapeutic Target for Recovery after Stroke and Traumatic Brain Injury*, in *Cell*, 176, 5, 2019, 1143-1157; M. Zhou, S. Greenhill, S. Huang et al., *CCR5 is a Suppressor for Cortical Plasticity and Hippocampal Learning and Memory*, in *eLife,* 2016.

[6] L.B. Andrews, N. Elster, *Regulating Reproductive Technologies*, in *Journal of Legal Medicine,* 21, 1, 2000, 35-65; L.M. Silver, *Remaking Eden. How Genetic Engineering and Cloning Will Transform the American Family*, New York, 1998; S.M. Suter, *A Brave New World of Designer Babies?,* in *Berkeley Technology Law Journal,* 22, 2007, 897-960; S.M Suter, *The Tyranny of Choice: Reproductive Selection in the Future,* in *Journal of Law and the Biosciences,* 5, 2, 2018, 262-300.

[7] J. Robertson, *Children of Choice: Freedom and the New Reproductive Technologies*, Princeton, N.J., 1994; J. Robertson, *Genetic Selection of Offspring Characteristics*, in *Boston University Law Review,* 76, 1996, 421-482; J. Robertson, *Extending Preimplantation Genetic Diagnosis: Medical and Non-Medical Uses*, in *BMJ: Journal of Medical Ethics,* 29, 2003, 213-216.

ISSN 2284-4503

arguing that parental decisions are supported by the figure of reproductive rights. The paper will argue that reproductive rights do not include NHRRD, but merely include in their scope parental reproductive decisions related to the health of progeny. Based on this assumption, the paper will conclude that to select features related to health is a choice grounded on reproductive rights, but all other types of reproductive parental choices cannot find proper legal and ethical ground in the norms granting reproductive rights (part 4).

However, this conclusion does not close the discussion. This would equate to sustain that conducts are generally prohibited unless they count with arguments in favour, when in fact it is the opposite: conducts are generally allowed unless there are sound arguments leading to prohibition. Even though NHRRD cannot find proper justification in reproductive rights, there are no sound arguments against NHRRD that justify their legal prohibition. Furthermore, the prohibition is very difficult to enforce because it faces two major difficulties. First, it assumes that there is a clear distinction between features related to health and all the remaining ones, which is not the case. Secondly, it assumes that characteristics not related to the health of the prospective child are irrelevant for his/her life, nothing more than parental whims or caprices, when in fact some of them can be as decisive as health-related features (part 5).

This paper argues that the traditional generalized ban on NHRRD should be reassessed and that some forms of NHRRD should be allowed, depending on the specific medical/scientific mechanisms used and the specific traits being selected (part 6).

2. Parental reproductive choices in light of the existing regulations

Parental reproductive choices and genetic planning are not allowed without limitations, which vary from jurisdiction to jurisdiction.

In what regards abortion, most jurisdictions in the world accept it when the aim is to prevent the birth of child with a severe disease or disability (though eventually conditioned by a temporal requisite).[8] In contrast, abortion based on non-health related characteristics of the child is generally banned.[9] In particular, abortion based on race/ethnicity[10] or gender[11] violates anti-discrimination laws. This trend might also be extended (under the same accusation of discrimination) to genetic abnormalities,[12] a classic justification for lawful abortion, that continues to be generally accepted in almost all

[8] A. GUILLAUME, C. Rossier, *L'avortement dans le Monde. État des Lieux des Législations, Mesures, Tendances et Consequences*, in *Population*, 73, 2, 2018, 217-306.
See also https://maps.reproductiverights.org/worldabortionlaws (last visited 02/07/2020).

[9] Note that the so-called abortion on request – abortion based on the sole request of the woman, without specifying the motivation, usually within a legally established time frame – might lead to that result, since the desire to avoid a certain feature of the child might be the woman's real motivation.

[10] E. GREEN, *Should Women Be Able to Abort a Fetus Just Because It's Female?,* in *The Atlantic*, May 16, 2016, at https://www.theatlantic.com/politics/archive/2016/05/sex-disability-race-selective-abortion-indiana/482856/ (last visited 06/06/2020).

[11] S. ANITHA and A.K. GILL, *Making Politics Visible: Discourses on Gender and Race in the Problematisation of Sex-Selective Abortion*, in *Feminist Review,* 120, 1, 2018, 1-19; E. GREEN, *op. cit.*; E. LEE, *Constructing Abortion as a Social Problem: 'Sex Selection' and the British Abortion Debate*, in *Feminism & Psychology*, 27, 1, 2017, 15-33.

[12] E. GREEN, *op. cit.*

jurisdictions of the EU[13] (except for Malta, that bans abortion in every circumstance).[14] However, since the parent's motivation is usually not disclosed, there is no chance to prevent it.

The same limit is present in reproductive techniques and associated procedures. PGD is a common mechanism for parents to exercise their reproductive choices, but in Europe those choices tend to be restricted to health-related characteristics.[15] The same is valid for gamete donation. The users of reproductive techniques can request the gametes of third parties (the donors) if the prospective parents are unable to procreate (infertility, reproduction by a single woman, gay couples) or if there is the risk of transmitting a serious medical condition to offspring.[16] However, prospective parents cannot resort to gamete selection to have a child with specific desired features. The selection of a donor having as criterion his/her facial traits, IQ or sports talent is banned in Europe. The only feature in which parents have a word to say regards phenotypic traits aimed to emulate the parents' phenotype and to create the fiction of a genetically based family. Even though parents cannot directly select children that look like them, some laws and/or medical guidelines demand the selection of donors that share their same phenotype, namely their ethnic group.[17] In Europe, this restriction is based on basilar principles coming from the Convention on Human Rights and Biomedicine.[18] Article 14 very clearly states: "The use of techniques of medically assisted procreation shall not be allowed for the purpose of choosing a future child's sex, except where serious hereditary sex-related disease is to be avoided".

Gene editing is not yet a common practice for parents to exercise their reproductive choices, even though it might become so in the future. The general trend in European laws is to allow genetic interventions aimed to cure or prevent diseases as long as their effects are restricted to the person himself/herself. Genetic ameliorations not related to health are forbidden. The Convention on Human Rights and Biomedicine also follows this solution.[19] According to its Article 13: "An intervention seeking to modify the human genome may only be undertaken for preventive, diagnostic or therapeutic purposes and only if its aim is not to introduce any modification in the genome of any descendants".

In conclusion, in Europe (the same is valid for several other jurisdictions around the world, the US being an exception in some ways) parental reproductive choices and genetic planning are accepted (*rectius*,

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[13] Associazione Luca Coscioni per la Libertà di Ricerca Scientifica, *Interruzione volontaria di gravidanza nei Paesi dell'Unione Europea,* 2018, at https://www.associazionelucacoscioni.it/wp-content/uploads/2016/09/IVG-nellUnione-Europea-1.pdf (last visited 04/08/2020).

[14] M. Webb, *A Year Later, and Abortion Is Not Yet Legal in Malta,* in *Dalli, Newsbook*, June 26 2019, at https://www.newsbook.com.mt/artikli/2019/06/26/a-year-later-and-abortion-is-not-yet-legal-in-malta-dalli/?lang=en (last visited 20/05/2020).

[15] M.J. Bayefsky, *Comparative Preimplantation Genetic Diagnosis Policy in Europe and the USA and its Implications for Reproductive Tourism*, in *Reproductive Biomedicine & Society Online*, 3, 2016, 41-47; T. Lemke, J. Rüppel, *Social Dimensions of Preimplantation Genetic Diagnosis: A Literature Review*, in *New Genetics and Society,* 38, 1, 2019, 83-84.

[16] M. Richards, G. Pennings, J.B. Appleby (eds), *Reproductive Donation: Practice, Policy, and Bioethics*, Cambridge, UK, 2012.

[17] In detail about this issue see V.L. Raposo, *Wrongful Genetic Connection: Neither Blood of My Blood, nor Flesh of My Flesh*, in *Med Health Care and Philos*, 23, 2019, 318.

[18] Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine (4 April 1997).

[19] F. Baylis, L. Ikemoto, *The Council of Europe and the Prohibition on Human Germline Genome Editing,* in *EMBO Rep,* 18, 2017, 2084-2085.

generally accepted, since even in this regard there are some exceptions) when they involve health-related features but banned in the remaining cases.

3. Classic arguments against NHRRD

The classic arguments to ban NHRRD can be summarized as such: eugenics, loss of genetic diversity, genetic discrimination, degradation of the child to become an object of a parent's wishes, limitations on the child's free development and replacement of the natural order of things by an artificial human order. This section will demonstrate there is not a sound argument to forbid NHRRD.[20]

3.1. Eugenics

Eugenics is probably the most invoked arguments against NHRRD, based on the assimilation between the latter and some atrocities that happened in the past.[21] The qualification of NHRRD as eugenics depends on the definition of this practice. In a broad sense NHRRD can be considered a form of eugenics because they do intent to select "better" features for prospective human beings.
However, what exact features are considered "better" is open to discussion. In the type of eugenics (assuming it is indeed eugenics, a kind of new eugenics,[22] also called liberal eugenics)[23] we have nowadays, features are picked by parents according to their subjective preferences, so, they might or might not be objectively an enhancement. Parents personal preferences do not necessarily lead to the amelioration of offspring, even though those particular parents consider it to be an amelioration.[24] For instance, some parents might prefer a child with blue eyes, but this eye colour is not necessarily praised as more valuable by the remaining community. Eventually, a genetic selection that parents consider

[20] For a critic to these arguments, F. ALLHOFF, *Germ-Line Genetic Enhancement and Rawlsian Primary Goods,* in *Kennedy Institute of Ethics Journal*, 15, 1, 2005, 39-56; G. BOGNAR, *Enhancement and Equality*, in *Ethical Perspectives*, 19, 1, 2012, 11-32; C. GYNGELL, T. DOUGLAS, J. SAVULESCU, *The Ethics of Germline Gene Editing*, in *Journal of Applied Philosophy*, 34, 4, 2017, 498-513; D.B. RESNIK, D.B. VORHAUS*, Genetic Modification and Genetic Determinism,* in *Philosophy, Ethics, and Humanities in Medicine*, Article ID 9, 2006.

[21] L. KASS, *Life, Liberty and the Defense of Dignity*, San Francisco, 2002; M.J. SANDEL, *The Case Against Perfection,* in *The Atlantic*, April 2004, at https://www.theatlantic.com/magazine/archive/2004/04/the-case-against-perfection/302927/ (last visited 06/06/2020); S.M. SUTER, *A Brave New World,* cit.*;* S.M. SUTER, *The Tyranny of Choice,* cit.

[22] J. DAAR, *The New Eugenics: Selective Breeding in an Era of Reproductive Technologies*, New Haven, Connecticut, 2017; M.J. SELGELID, *Moderate Eugenics and Human Enhancement,* in *Medicine, Health Care, and Philosophy,* 17, 1, 2014, 6-8.

[23] In favour of liberal eugenics, for instance, E. FENTON, *Liberal Eugenics and Human Nature. Against Habermas*, in *Hastings Cent Rep*, 36, 6, 2006, 35-42; D. FOX, *The Illiberality of 'Liberal Eugenics',* in *Ratio,* 20, 2007, at https://papers.ssrn.com/sol3/papers.cfm?abstract_id=1072104 (last visited 03/04/2020).
These concepts – private eugenics or liberal eugenics – should not be confused with the new eugenics referred by Judith Daar in one of her latest books (J. DAAR, *The New Eugenics, cit*.) to refer the denial of reproductive technologies to some population strata, such as racial minorities, persons with disabilities, gay unions, and single people.

[24] S.M. SUTER (*A Brave New World,* cit.*,* 934 ff.) seems to consider that all non-health-related features are directed to enhancement, but this is not exactly so, or at least not all of them aim an objective enhancement, i.e., some features might be considered by parents as an amelioration but by other people as a neutral feature.

an improvement for the child might even be in objective terms a harm,[25] as it happens when parents deliberately select a deft child.[26]

Even if we agree that both of them are eugenics practices, current private eugenics is totally different from past public eugenics in their ethical/philosophical base, their *modus operandi* and their goals.[27] The experiences we had in the past – whose apogee was the Nazism – were based on the assumption that some human beings had higher value than the others and thus that these others should be eliminated in the name of racial purity.[28] Based on this experience of dehumanization of human beings[29] the concept was demonised.

In contrast, the kind of eugenics we have today is not related with the intention of purifying or improving humankind, but merely with parental decisions on what they believe to be better for their children.[30] Even if NHRRD are considered eugenics, it must be recognised that this is a totally different kind of eugenics and that the (very powerful) arguments against traditional eugenics are ungrounded regarding NHRRD. There are four main difference between NHRRD and traditional eugenics i) While traditional eugenics was guided (*rectius*, imposed) by the State, NHRRD consist in purely private decisions; ii) The motivations of NHRRD are very different from the motivations of traditional public eugenics, as its aims is to satisfy parents' wishes and their vision of what is best for the child; iii) Within NHRRD the target is a particular child while in traditional eugenics the target was the humankind in general; iv) Moreover, the features desired for humankind probably do not coincide with the ones that parents envisage for their children, since parents take their reproductive and genetic decisions based on personal and very subjective assessments, which do not always lead to the objective amelioration of the child.

[25] About these cases see V.L. RAPOSO, *The Usual Suspects: Can Parents Be Held Accountable for Their Reproductive And Genetic Decisions*?, in *Revista de Derecho y Genoma Humano: Genética, Biotecnología y Medicina Avanzada/Law and the Human Genome Review: Genetics, Biotechnology and Advanced Medicine,* 47, 2017, 109-137.

[26] M. SPRIGGS, *Lesbian Couple Create a Child Who Is Deaf Like Them*, in *Journal of Medical Ethics,* 28, 2002, 283. Regardless of the controversy about the qualification of deafness as a disability (which will not be developed in this paper), this preference was grounded on their subjective personal conditions, not on an objective positive assessment of deafness.

[27] The difference between traditional public eugenics and private eugenics in G. CAVALIERE, *Looking into the Shadow: The Eugenics Argument in Debates on Reproductive Technologies and Practices*, in *Monash Bioethics Review*, 36, 1-4, 2018a, 1–22; T. DOUGLAS, K. DEVOLDER, *Procreative Altruism: Beyond Individualism in Reproductive Selection,* in *The Journal of Medicine and Philosophy,* 38, 4, 2013, 400–419; V.L. RAPOSO, *Gene Editing, the Mystic Threat to Human Dignity*, in *Journal of Bioethical Inquiry*, 16, 2, 2019, 249-257.

[28] Nazism, and traditional eugenics in general, is much more complex that this brief description, which merely highlights its basic features. Further details in S. GRAUMAN, *Germline Gene Therapy: Public Opinions with Regard to Eugenics*, in E. Hildt, S. Graumann, (eds.), *Genetics in Human Reproduction*, Aldershot, 1999, 175; M. MALINOKWSI, *Choosing the Genetic Makeup of Children: Our Eugenics Past-Present, and Future,* in *Connecticut Law Review,* 36, 1, 2003, 134 ff.; S.M. SUTER, *A Brave New World,* cit., 901 ff.

[29] Traditional public eugenics certainly was (S. BACHRUCH*, In the Name of Public Health - Nazi Racial Hygiene*, in *New England Journal of Medicine* 351.5, 2004, 417– 420; V. FINKELSTEIN, O. STUART, *Developing New Services,* in G. Hales (ed.), *Beyond Disability: Towards an Enabling Society*, London, 1996, 170-187.

[30] In favour of liberal eugenics, for instance, E. FENTON, *op. cit*; D. FOX, *The Illiberality of 'Liberal Eugenics',* cit.

3.2. Loss of genetic diversity

One of the criticisms of NHRRD has been that human beings will all become the same, like products leaving an assembly line.[31] The argument claims that the loss of genetic diversity[32] and of "social heterosis"[33] will undermine the very survival of the human species. The existence of a plurality of genes makes humankind stronger to face challenges posed by virus, bacteria, and even climatic and environmental changes,[34] because it equips the human species with genetic resources to adapt to these events.

However, chances are that parents will not select the exact same characteristics for their offspring. Some may prefer children with blond hair, and others may prefer dark hair; some may prefer boys, and others may prefer girls. It is a fact that some features are more likely to be picked than others, taking into consideration what society considers to be beautiful or desirable. In a society that glorifies beauty, intelligence and physical power it can be assumed that these traits will be the most selected, provided science allows it. Nonetheless, the concept of beauty remains subjective, and so the choices will hardly coincide.

3.3. Genetic discrimination

The peril of discrimination has also been frequently invoked (genetic discrimination) against NHRRD.[35] The accusation of discrimination is based in two different (though related) arguments: i) money as a barrier to access NHRRD; ii) and the consequent prejudice against the ones that have not benefit from NHRRD.

The first argument states that NHRRD are discriminatory because only the ones with sufficient financial power can have the chance to decide the features of their offspring and consequently only their children can be improved (assuming that indeed these decisions will improve them) by NHRRD.[36] It will be money, and not merit or luck, to determinate the ones that have better chances in life. Let us take the case of intelligence: today there are few geniuses and thus they are praised; but in a genetically selected/modified society anyone whose parents can pay for fancy genetic techniques can become a genius, for the simple reason that they come from a wealthy family.

However, in many ways money already dictates one's fate. Parents with money can offer a better education to their children, provide better health care, give them various opportunities (universities, travels) and buy things that can make them look more attractive and eventually more socially accepted (fancy clothes, cosmetic interventions). So, the possession of wealth (or the lack of it) can indeed condition the possibilities open to someone. It can be said that all possibilities that depend on money

[31] P. UNALKAT, *Alubias, Genes y Temas*, in C. Romeo Casabona (ed.), *La Necesidad de la Precaución, in Biotecnología y Derecho: Perspectivas en Derecho Comparado,* Bilbao-Granada, 1998, 399-404.

[32] P.H. HUANG, *Herd Behavior in Designer Genes*, in *Wake Forest Law Review*, 34, 3, 1999, 645-653.

[33] P. NONACS, K.M. KAPHEIM, *Social Heterosis and the Maintenance of Genetic Diversity*, in *Journal of Evolutionary Biology*, 20, 6, 2007, 2253-2265.

[34] D.B. RESNIK, *Of Maize and Men: Reproductive Control and the Threat to Genetic Diversity*, in *Journal of Medicine and Philosophy*, 25, 4, 2000, 451-451.

[35] R. RAO, *Equal liberty: Assisted Reproductive Technology and Reproductive Equality*, in *George Washington Law Review*, 78, 2008, 1467; S.M. SUTER, *A Brave New World,* cit., 940 ff., 954 ff.

[36] M.A. ROTHSTEIN, *Legal Conceptions of Equality in the Genomic Age,* in *Law & Inequality,* 25, 2007, 429-463.

are to be repudiated and only possibilities conditioned by luck (that is, genetic lottery) should be accepted, but it remains to be seen why luck is better (more legitimate, fairer) than wealth.
From a different perspective, it has been stated that the referred inequalities should also be prevented,[37] so, they cannot serve as justification to other inequalities.[38] But if the aim is to combat inequality, why should we focus only on the ones based on money and the possibilities it can provide? What about the ones based on genetic dotation? The genotype and the phenotype of individuals is not all the same: some are healthy than others, some are more intelligent than others, some are more fit than others. Benefits based on money are not morally better than benefits based on luck. From this perspective, NHRRD could actually create some biologic equality among us.
The second argument is grounded on the previous one: since not everyone can beneficiate from NHRRD, the ones that are deprived of that possibility will be ostracised and thus this practice shall be forbidden. Taking again the example of intelligence, the children whose parents did not select a higher IQ will be considered less than the ones that were made "more intelligent". However, no matter how genetically homogeneous a society is, people with different features will always exist and they might be discriminated against.[39] The only way to avoid this outcome would be to create a society where everyone is exactly the same, but that scenario would certainly be criticized based on the argument of loss of genetic diversity.

3.4. Objectification of children

The risk of a child being objectified has also been argued (child instrumentalization).[40] In the United States, it is common to find in newspapers advertisements looking for gamete donors (male or female) with certain desired characteristics, as "products" for sale.
A "tailor-made child", chosen like a supermarket product, with a certain hair or eye colour, must be rejected. However, the argument of objectification lies in a profoundly naïf conception of parenthood, according to which parents raise their children as a blank canvas. This is obviously not the case. Parents use several mechanisms (apart from the genes) to create the kind of chid they wish.[41] The principles they teach to their children, the school they pick, the option between piano lessons or swimming lessons, all these conditions the kind of person the child will be.[42]

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[37] M.J. SELGELID, *op. cit.,* 10-11. About State duties to combat inequalities see F. GARCÍA GIBSON, *Conflicts Between Domestic Inequality and Global Poverty: Lexicality Versus Proportionality*, in *Ethics & Global Politics*, 9,1, 2016.
[38] E. WESLEY, F. PETERSON, *Is Economic Inequality Really a Problem? A Review of the Arguments*, in *Soc. Sci*., 147, 2017.
[39] H. LOU, *Eugenics Then and Now: Constitutional Limits on the Use of Reproductive Screening Technologies*, in *Hastings Constitutional Law Quarterly*, 42, 2015, 409 (arguing the diminution of tolerance against those who are different).
[40] For criticism about the child's instrumentalisation, see D.W. JORDAAN, *Preimplantation Genetic Screening and Selection: An Ethical Analysis*, in *Biotechnology Law Report*, 22, 6, 2003, 589; S.M. SUTER, *A Brave New World*, cit., 960 ff.
[41] Parents do so in compliance with the parental duties imposed by family law (cf. K. BOELE-WOELKI, F. FERRAND, C. GONZÁLEZ-BEILFUSS, M. JÄNTERÄ-JAREBORG, N. LOWE, D. MARTINY,W. PINTENS, *Principles of European Family Law Regarding Parental Responsibilities*, Antwerpen, 2007).
[42] The analogy between environmental and social determinism and genetic determinism in N. AGAR, *Liberal Eugenics*, in H. KUHSE, P. SINGER (eds.), *Bioethics: An anthology*, Oxford, U.K., 1999, 172-173; A. BUCHANAN, D.W.

The fact that parents try to shape children according to their own wishes[43] (although usually having also in consideration the children's best interest) demonstrates that people do not have children for the child's sake, bur for their own sake. The goal that motivates reproduction is a very selfish one: to create a human being that parents can model in light of one ideal.[44] Parents are willing to fully accept their children and love them, but still they try to select a child with features they highly praise.[45]
It can be argued that the two types of conditioning – the environmental and the genetic - are very different and that they cannot be compared in their *modus operandi*, and above all in their consequences. Quoting Prusak, « [w]hereas environmental manipulations work on phenotype, prenatal genetic manipulations would work, of course, on genotype».[46] Based on this distinction, the author states that though children can overcome whatever expectations parents could have created when picking a specific upbringing, this is not possible regarding the expectations they created by means of NHRRD.[47] But if the discussion is based on parents' expectations (similar to Habermas' argument on intentions that will be exposed below), the fact is that such expectations exist in both cases. Some parents that send their child at a young age to music classes expect to raise a musician, and if they don't achieve that goal, they will be as disappointed as the ones that resorted to NHRRD. Moreover, Prusack's statement that environmental conditioning only affects the phenotype, and for that reason this type of conditioning does not raise problems, might lead to the impression that phenotypes are less decisive than genotypes, which is not accurate.[48] The phenotype can dictate characteristics easily perceived by the community to praise or repudiate, and therefore dictate our chances in that community. Moreover, environmental conditioning can shape features to be passed to the offspring (skin tone, eye colour)[49] and therefore condition an entire lineage.

3.5. Violation of the child's right to an open future

It has been said that the NHRRD imposes on the child a certain destiny, that the child cannot modify, and thus he/she is forced to live the life that parents picked for him/her. Due to limitations on his/her

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BROCK, N. DANIELS et al., *From Chance to Choice: Genetics and Justice,* Cambridge, 2000, 160-161; J. ROBERTSON, *Children of Choice,* cit.,167.

[43] See D. BENATAR, D. WASSERMAN, *Debating Procreation: Is It Wrong to Reproduce?,* New York, 2015, 183.

[44] V.L. RAPOSO, *O Direito à Imortalidade (O Exercício de Direitos Reprodutivos Mediante Técnicas de Reprodução Assistida e o Estatuto Jurídico do Embrião In Vitro,* Coimbra, 2014, 250, 351.

[45] J. MALEK, *Use or Refuse Reproductive Genetic Technologies: Which Would a 'Good Parent' Do*?, in *Bioethics,* 27, 2, 2013, 63-64.

[46] B.G. PRUSAK, *Rethinking 'Liberal Eugenics': Reflections and Questions on Habermas on Bioethics*, in *Hastings Cent Rep,* 35, 6, 2005, 38. In the same sense, J. HABERMAS, *The Future of Human Nature,* Cambridge, 2003, 61.

[47] B.G: PRUSAK, *op. cit.,*38. In the same sense, J. HABERMAS, *op. cit.,*61.

[48] The differences between the two in P. TAYLOR, R. LEWONTIN, *The Genotype/Phenotype Distinction*, in Edward N. Zalta (ed.), *The Stanford Encyclopedia of Philosophy* (Summer 2017 Edition), https://plato.stanford.edu/archives/sum2017/entries/genotype-phenotype/ (last visited 05/06/2020).

[49] R.A. STURM, D.L. DUFFY, *Human Pigmentation Genes Under Environmental Selection*, in *Genome Biology*, 13, 9, 2012.

free development, the child could never be the master of his/her own destiny. The conclusion would be that NHRRD violate the child's right to an open future[50].

Habermas, for instance, argues that a person whose genes have been selected by third parties (assumingly, the parents) will never feel himself/herself genuinely free, as the true author of his/her life[51]. Or, using Sandel's words: "children are free to choose their characteristics for themselves. But none of us chooses his genetic inheritance. The alternative to a cloned or genetically enhanced child is not one whose future is unbound by particular talents but one at the mercy of the genetic lottery".[52]

However, both arguments (this one and the previous) rely on a premise that science proved to be wrong: genetic determinism, that is, the idea that genes would rigidly determine all our traits, without any possibility for surprise or divergence.

As Resnik and Vorhaus[53] very accurately demonstrated, genetic determinism is totally misleading. The success of any selection varies according to environmental influences. For instance, if parents select a physical trait, such as hair, skin or eye colour, they have more control over the result because these features are exclusively determined by genes, even though they are epistatic, (i.e., they do not depend on a single gene) and therefore difficult to "control". Alternatively, if parents look for an embryo with a higher level of intelligence, they can never be sure the child will effectively be more intelligent because environment (education, experience) is as decisive, or even more influential, than genetics, turning intelligence into a complex trait. Furthermore, epigenesis (the development of an embryo, starting from the single cell stage) plays a decisive role in the way genes express themselves; therefore, two people with the same exact genetic dotation (twins, clones) can become very different due to environmental influences and the individual development of the organism. Even though they share the exact same genotype, their features, their preferences and their skills might be completely different.[54]

If this is so, genetic selection does not turn children into parental "projects" whose fate is already determined. Parents cannot expect more from a baby with a "musical gene" than from a toddler who attended a couple of music classes. In both cases parents have simply provided the child with some tools for musical talent, but the final result is absolutely unpredictable.[55] Simultaneously, the child is not condemned to be a Mozart. The child might turn out to be a football player or an avid reader, without any particular musical talent. A natural (or, in this case, not that natural) gift for a certain

[50] J. FEINBERG, *The Child's Right to An Open Future*, in W. Aiken, H. LaFollette (eds.), *Whose Child?,* Totowa, NJ, 1980, 124-153. This argument is further developed by D. DAVIS, *Genetic Dilemmas and the Child's Right to an Open Future*, in *Rutgers Law Journal*, 28, 2, 1997, 549-592.

[51] J. HABERMAS, *op. cit.,* 25

[52] M.J. SANDEL, *op. cit.*

[53] D.B. RESNIK, VORHAUS, *op. cit.*

[54] R. PLOMIN, N.G. SHAKESHAFT, A. MCMILLAN et al., *Nature, Nurture, and Expertise*, in *Intelligence*, 45, 2014, 46-59.

[55] For more on the excessive pressure on parents over these children and the consequential failure of expectations, see L.A. VACCO, *Preimplantation Genetic Diagnosis: From Preventing Genetic Disease to Customizing Children. Can the Technology Be Regulated Based on the Parents' Intent?,* in *Saint Louis University Law Journal,* 49, 2005, 1195-1196.

activity does not dictate one's future, nor even one's ability to perform that activity, unless it has been properly nurtured and developed.[56]

Furthermore, even disregarding the possibility to decide the genetic code of their offspring, parents have many ways to define the child's life, by living in a certain place (a big city or a small rural village), selecting a certain school, and especially by the kind of education and values they transmit in their upbringing.[57] NHRRD are merely a mechanism, and by far not the most decisive, to shape the child's future.

Habermas tries to destroy the argument of genetic determinism by resorting to the underlying intention: it does not matter if genes can actually condition our life, but only the intention underlying the selection of those genes.[58] Transposing this reasoning to our questions: it is irrelevant how effective NHRRD can actually be, what is relevant is the purpose parents had in mind in their NHRRD. However, this argument if confusing. It is not clear what Habermas means by "intention". The logical answer is the parent's purpose, but when explaining the argument the author states that: "[t]he person concerned knows that the manipulation has been carried out with the sole intention of acting on the phenotype…". Therefore, one must wonder to which intention is Habermas referring to: the parents' real intention or what the children believe was the parents' intention? Whatever the answer ends up being, there is still a shortcoming in this argument. For it to be correct, it would have to be demonstrated that when educating their children parents do not desire to actually shape the child (or children would have to believe that parents do not actually desire that), while in NHRRD they effectively want to do so (or children believe they effectively want to do so). This is an assumption still waiting to be confirmed. Moreover, it is ambiguous (to say the least) to ground an argument on intentions or on what other believe those intentions to be.

3.6. The artificial over the natural

Let's assume, for the sake of the argument, that genetic determinism is correct. If that is the case, then, NHRRD would be as much a violation of the child's right to an open future as (coital) reproduction itself. The mere fact that parents (involuntary) pass their genes to offspring conditions the child life: if parents have a propensity to be fat probably the child will also have it,[59] what might prevent him/her from being a sport star or a model; if parents have Attention Deficit Hyperactivity Disorder children have 50% chances of inheriting it,[60] which will obviously have a huge impact on the child's life and on his/her future as an adult. So, in a way, parents always condition the kind of life and possibilities their children will have. The difference is that this conditioning is considered natural, and therefore good, while the conditioning carried out by NHRRD is considered artificial and therefore bad.

[56] For a detailed exposure of the failures of genetic determinism, see D.B. RESNIK, D.B. VORHAUS, *op. cit.* See also S.M. SUTER, *A Brave New World*, cit., 939-940.

[57] N. AGAR, *op. cit.*, 172-173; E. FENTON, *op. cit.*, 38.

[58] J. HABERMAS, *op. cit.*, 124.

[59] V.V. THAKER, *Genetic and Epigenetic Causes of Obesity*, in *Adolescent Medicine: State of the Art Reviews,* 28, 2, 2017, 379-405.

[60] 'About 40% of ADHD children have at least one parent with clinical ADHD symptoms' (M. STARCK, J. GRÜNWALD, A.A. SCHLARB, *Occurrence of ADHD in Parents of ADHD Children in a Clinical Sample*, in *Neuropsychiatr Dis Treat*., 12, 2016, 586).

Mainstream scholars share a fascination over the natural order of things.[61] This allure faces two problems. First, the difficulty in distinguishing what is natural to what is artificial, because "genetic technologies threaten this distinction because they enable manipulation of one subject by another and thereby blur the line between what is grown (the natural) and what is made (the artificial)".[62]
Secondly, the lack of justification of why natural is better.[63] Many things that happen in nature, without any type of human intervention, cannot be considered good by any parameter: young babies that die in their sleep, earthquakes and other natural disasters that kill millions. The lack of accountability, legal or moral, does not make this natural event good, or even neutral.
On the other hand, most aspects of human life nowadays are artificial (in the sense they are manmade), but still very beneficial to humankind. The fact that we, humans, have developed medicine to treat severe diseases and safe lives, for instance. This is far from natural. Under the rule of nature people with tuberculosis, pneumonia or even flu would die. I wonder if those scholars would find their death valuable just because imposed by nature.

4. Parental reproductive choices in light of reproductive rights

Mainstream scholars sustain that reproductive rights[64] cannot justify the power to decide whether a baby is going to be blond or brunette, male or female. [65] Any of these choices would constitute a reproductive rights abuse.[66]
I agree with the thesis that reproductive rights do not include in their scope the possibility to determine offspring characteristics not related with health.[67] When someone decides to have a child, there are some expectations that should be preserved. The decision to procreate involves burdens that parents implicitly accept, related to raising, educating and taking care of their child. There is no exact measure or quantity of burdens, but a commonly shared idea accepts there is a reasonable level of parental obligations that "come with the job". When dealing with a severely ill child, parents bear an enormous

[61] For instance, Habermas claims in favour of a "right to a genetic inheritance immune from artificial intervention" (J. HABERMAS, *op. cit.,* p. 27); Annas considers many of the practices included in NHRRD as crimes against humanity because they undermine human nature (G.J. ANNAS, *American Bioethics: Crossing Human Rights and Health Law Boundaries,* Oxford, U.K., 2005, 37).
[62] E. FENTON, *op. cit.,* 38.
[63] A similar critique, for the specific case of gene editing, in I. DE MIGUEL BERIAIN, L. MASTRANGELO, *Cosa c'è di Sbagliato nel Modificare la Linea Germinale?,* in *BioLaw Journal – Rivista di BioDiritto*, 1, 2020, 240-241.
[64] Some authors use the concept "reproductive rights" (V. ROZÉE GOMEZ, S. UNISA, *Surrogacy from a Reproductive Rights Perspective: The Case of India, in Autrepart,* 70, 2, 2014, 185-203), while others prefer "reproductive liberties" (T. BALDWIN, *Reproductive Liberty and Elitist Contempt: Reply to John Harris*, in *Journal of Medical Ethics*, 31, 5, 2005, 288-290). This dichotomy will not be discussed in the paper.
[65] Cf. S.M. SUTER, *A Brave New World,* cit.; S.M. SUTER, *The Tyranny of Choice,* cit.
In opposite sense, J. ROBERTSON, *Genetic selection*, cit., 424-432; J. HARRIS, *Rights and Reproductive Choice*, in J. HARRIS, S. HOLM (eds.), *The Future of Human Reproduction.* Oxford: 1998, 34; J. ROBERTSON, *Assisted Reproduction, Choosing Genes, and the Scope of Reproductive Freedom,* in *George Washington Law Review,* 76, 2008, 1490-1512.
[66] This paper will not discuss the existence of reproductive rights (for this see J. HARRIS, *Rights and Reproductive Choice,* cit.).
[67] V.L. RAPOSO, *The Usual Suspects,* cit.

responsibility, that foes beyond what is reasonably expected. Therefore, no parent should be forced to keep a child with a severe disability, unless clear and informed consent has been given. Future parents should demand to be informed about the health hazards of their future children by using different methods of preconception diagnosis. If they decide to go forward, they should be informed about reproductive techniques and associated genetic interventions to eradicate severe medical conditions. Parents should not be forced to procreate if they know their child would be severely disabled. Likewise, if they are aware of a genetic predisposition to a certain disease, they should be allowed to employ scientific procedures (reproductive treatments using gamete donation, PGD, genetic engineering) to avoid passing that predisposition on to their offspring. This parental right is concomitantly a duty towards offspring, as with many other rights belonging to parents in the relationship with their children. Therefore, not only are we talking about a right belonging to those who want to reproduce (reproductive right), but also to a duty imposed on these individuals to protect their future children (reproductive duty).[68] "We do not have a moral duty to bring people into existence with good lives; but we do have a moral duty to prevent the existence of people who would experience so much pain and suffering as to make their lives not worth living for them on the whole".[69]

Health is a relevant consideration and having a healthy child can be a decisive factor in the decision to reproduce. Some non-health related features can also play the same role.[70] In this regard John Roberson provided the following example: [71] for a family dedicated to music, musical talent can be a decisive condition to reproduce, even though this is a mere subjective preference. However, for that family it can be as decisive as having a healthy child, which is an objective and universal preference.[72] However, this preference is not protected by reproductive rights.

Parents have a right (as an aspect of the right to reproduce) to ensure that their child will not suffer from a certain disease[73] (even this dimension is contested by some authors)[74] and to use scientific mechanisms to achieve that objective,[75] grounded on their reproductive rights. However, reproductive rights do not provide parents the possibility to determine other types of characteristics for their

[68] The nature of such duty – legal or moral – will not be discussed here. For further developments see V.L. RAPOSO, *The Usual Suspects,* cit.*;* V.L. RAPOSO, Bons Pais, *Bons Genes?: Deveres Reprodutivos no Domínio da Saúde e Procreative Beneficence*, in *Lex Medicinae,* 4, II, 2019, 471-483.

[69] W. GLANNON, *Genes, Embryos, and Future People*, in *Bioethics,* 12, 3, 1998), 188. See also J. HAMMOND, *Genetic Engineering to Avoid Genetic Neglect: From Chance to Responsibility*, in *Bioethics,* 24, 4, 2010, 160-169.
This dimension won't be addressed in this paper. More developments on this issue are found in J. HAMMOND, *op. cit.,* 160-169; V.L. RAPOSO, *The Usual Suspects,* cit.,109-237.

[70] C. GYNGELL, T. DOUGLAS, J. SAVULESCU, *op. cit.*

[71] J. ROBERTSON, *Extending preimplantation genetic diagnosis*, cit., 215.

[72] As an exception, there are cases of parents that deliberately selected a child with a disease or a disability. See, for instance the case of a couple that selected a child with Down syndrome (M. HEALY, *Fertility's New Frontier: Advanced Genetic Screening Could Help Lead to the Birth of a Healthy Baby*, in *L.A. Times*, July 21 2003, at https://www.latimes.com/archives/la-xpm-2003-jul-21-he-pgd21-story.html (last visited 06/04/2021).

[73] L.B. ANDREWS, N. ELSTER, *op. cit.,* 62 ff.

[74] J. MITTRA, *Marginalising 'Eugenic Anxiety' Through a Rhetoric of 'Liberal Choice': A Critique of the House of Commons Select Committee Report on Reproductive Technologies*, in *New Genetics and Society,* 26, 2, 2007, 159-179.

[75] V.L. RAPOSO, *O Direito à Imortalidade*, cit., 158 ff.

children.[76] As Lee Silver put it, «[t]here's a big difference between curing infertility, on the one hand, and trying to make sure that your child inherits your curly hair on the other».[77]

5. Problems faced by this position

According to the previous considerations, parents can select health-related features for their children but lack grounds to select other features. This conclusion, however, faces two major shortcomings. First, it does not provide a distinctive criterion for distinguishing health-related features from others. Second, it fails to demonstrate why health-related features are necessarily more relevant (that is, more decisive for the child's future) than non-health-related features.

5.1. The distinction between health-related features and other features

Doubts about NHRRD have arisen because there is no clear concept of health,[78] a doubt exacerbated by the World Health Organization's (too) broad definition of health as «a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity».[79] If we select a child whose characteristics can provide him or her with a happier existence and promote his or her well-being, such as having a sense of humour or empathy, this selection seems to amount to health-related features based on the WHO's definition and thus it would be allowed.

However, this is not the most commonly used operative concept of "health", eventually because of its extremely broad scope. Commonly, "health" appears related to medicine and medical decisions. This seems to be the distinctive criterion that grounds existing legal solutions, for instance, to define lawful abortion based on the unborn's features[80] and lawful PGD:[81] if intended to avoid the birth of a child with a disease or malformation, as such understood by medicine, these practices are allowed, if not they are banned.

However, this results in an additional problem. From a theoretical perspective, the distinction between health-related features (medically relevant choices) and non-health-related features (medically irrelevant choices) appears to be clear and justified on objective medical grounds. The problem is that

[76] Moreover, NHRRD are not included in the scope of protection of other rights (the more suitable candidates would be the right to privacy, the right to self-determination and the right to create a family). For this analysis see V.L. Raposo, *O Direito à Imortalidade*, cit., 157-169.

[77] L. SILVER, *op. cit.,* 75.

[78] Regarding this question, see J. ROBERTS, *Treating the Enhancement Debate: Irrelevant Distinctions in the Enhancement Medicine Debate,* in *Kriterion – Journal of Philosophy,* 28, 1, 2014, 1-12.

[79] WORLD HEALTH ORGANIZATION, *Preamble to the Constitution of the World Health Organization, as adopted by the International Health Conference,* New York, June 19-22, 1946, https://www.who.int/governance/eb/who_constitution_en.pdf (last visited 03/06/2020).

[80] Abortion Legislation in Europe, last update 12/30/2020, at https://www.loc.gov/law/help/abortion-legislation/europe.php (last visited 10 April 2021).

[81] A.M. DUGUET, B. BOYER-BEVIERE, *Preimplantation Genetic Diagnosis: The Situation in France and in Other European Countries*, in *Eur J Health Law*, 24, 2, 2017, 160-174.

in practical terms the difference is much more complex. What is a disease or a disability, as opposed to a condition not connected to health,[82] or how to distinguish treatment from enhancement?[83]
Not even an analogy with the scope of medicine helps in our definition because medicine has progressively expanded its scope to include plastic surgery, fertility procedures, sport medicine, and so on. If we select a child with certain facial characteristics or athletic ability, this selection is still within the scope of modern medicine and could be allowed under this criterion.
Furthermore, many believe that disease is a social construction,[84] that is, that classifying certain behaviour as a disease or not is socially determined. This formula opens the door to, for example, homosexuality or sex addiction being qualified as diseases because they are considered as such in certain communities. Under this (very contested) criterion, we can select a straight child or a child with controlled sexual behaviour[85] (assuming we could control features such as sexual orientation and sexual behaviour by means of genetics, a possibility that presently lacks a scientific basis) under the umbrella of health-related features.
As Sonia Suter puts it
on a societal scale, the more we use technology to select against lesser conditions and traits, the more perfectionist we may become as a culture, and the more demanding we may become with respect to what is acceptable, normal, or healthy.[86] In sum, the absence of a clear concept of "health" is a major drawback of mainstream theories that ban NHRRD.

5.2. The relevance of the selected features

Parents select certain features because they value them, i.e., those traits are not neutral for parents, even if the remaining members of the community do not consider them as valuable. The importance of this information is a subjective matter.[87] The specific content/design of the selected characteristics can vary widely because they basically depend on the subjective assessments of each set of parents. For instance, it is fair to say that most parents want a child physically attractive. However, the concept of beauty is far from uniform. Some may find a blond-haired child with brown eyes to be beautiful, whereas others may consider black hair and blue eyes to be more alluring.
Moreover, the apparent neutrality of non-health related features can be challenged. Some alleged "neutral characteristics" are indeed relevant to a child's future opportunities. For instance, in many

[82] Also questioning this distinction, N. AGAR, *op. cit*., 173-174.
[83] S. CHAN, J. HARRIS, *In Support of Human Enhancement,* in *Studies in Ethics, Law, and Technology,* 1, 1, 2007; J. HARRIS, *Enhancing Evolution*, Princeton, NJ, 2007; E. IGNOVSKA, G.F. BLASI, *Reproduction, the Key to Human Evolution: a Legal and Ethical Study*, in *BioLaw Journal – Rivista di BioDiritto*, 2, 2017, 117-119.
[84] Cf. P. CONRAD, K.K. BARKER, *The Social Construction of Illness: Key Insights and Policy Implications*, in *Journal of Health and Social Behavior,* 51, S, 2010, S67. For more on this issue see S.M. SQUIER, *Narrating Genetic Disabilities: Social Constructs, Medical Treatment, and Public Policy,* in *Issues in Law and Medicine,* 15,2, 1999, 141-158.
[85] Dahl, for instance, sustains that parents should be able to select the sexual orientation of their progeny. Cf. E. DAHL, *Ethical Issues in New Uses of Preimplantation Genetic Diagnosis*, in *Human Reproduction,* 18, 7, 2003, 1368-1369.
[86] S.M. SUTER, *A Brave New World,* cit.*,* 936.
[87] J. DAAR, *ART and the Search for Perfectionism: On Selecting Gender, Genes, and Gametes*, in *The Journal of Gender, Race and Justice,* 9, 2005, 269; J. ROBERTSON, *Genetic Selection*, cit.*,* 430.

societies tall people may be more highly valued, so parents may select embryos that produce taller human beings.[88] Some research has shown that appearance influences success. For instance, eye colour can condition your achievements in life ("blue-eyed people being more intelligent and brown-eyed people having faster reaction times").[89] Furthermore, not only is it common knowledge that attractive people have more success, but some studies have attested they generally have higher incomes.[90] What about sexual orientation? This is far from being a neutral feature. Most parents prefer "straight" children, primarily because they are more easily accepted by society.[91] Accordingly, one can assume that if a "gay gene" were to be discovered, some parents would select against it. In the case of gender, it can hardly be deemed a trivial feature when some cultures stigmatise females. Thus, even if gender selection is allowed in certain jurisdictions[92] disregarding health issues,[93] it should only be permitted in countries where male babies are not systematically desired, and females undervalued.[94] When prospective parents deem a feature to be important, in such a way that it conditions their decision to procreate (even if for the average person it is redundant), why not allow the selection of

[88] Some considerations about the value of being tall are discussed in C. GYNGELL, T. DOUGLAS, *Stocking the Genetic Supermarket: Reproductive Genetic Technologies and Collective Action Problems*, in *Bioethics*, 29, 4, 2015, 243.
However, if more tall people are born this may lead to two different and opposing consequences: the idea that being tall is better than being short might be perpetuated or being tall might become so common that being short will have more value.

[89] J. WATSON, *Eye Colour and Reaction Time: An Opportunity for Critical Statistical Reasoning*, in *Australian Mathematics Teacher,* 64, 3, 2008, 30-40.
See also P.J. ROWE, P. EVANS, *Ball Color, Eye Color, and a Reactive Motor Skill*, in *Perception Motor Skills,* 79, 1Pt2, 1994, 671-674.

[90] "The average cost of being ugly is $230,000 out of your paycheck over your working lifetime. Deduct another significant chunk from your salary if you are obese, but only if you are female. Fat women earn about $14,000 less per year than their average-weight sisters, or about 12% if you are Caucasian and 7% if you are African American. On the other hand, remarkably thin women earn $2,000 more each year than the average woman on the job" (N. ROSEN, *Blondes Really Do Earn More Money*, Sep. 1, 2011, http://www.businessinsider.com/the-ugly-tax-2011-8).

[91] B. WILLIAMS, *Screening for Children: Choice and Chance in the Wild West of Reproductive Medicine*, in *George Washington Law Review*, 79, 4, 2011, 1321-1322.

[92] In the absence of a legal ban, it is legally practiced in some US states, Ukraine, Thailand, Mexico.
In favour, B. STEINBOCK, *Sex Selection: Not Obviously Wrong*, in *Hastings Center Report,* 32, 1, 2002, 23-28; J. HARRIS, *No Sex Selection Please, We're British*, in *Journal of Medical Ethics*, 31, 5, 2005, 286-288.
Against, T. BALDWIN, *op. cit.*; R. MACDOUGALL, *Acting Parentally: An Argument Against Sex Selection*, in *Journal of Medical Ethics*, 31, 10, 2005, 601-605.
Presenting arguments for and against this possibility see the ETHICS COMMITTEE OF THE AMERICAN SOCIETY FOR REPRODUCTIVE MEDICINE, *Use of Reproductive Technology for Sex Selection for Nonmedical Reasons,* in *Fertility and Sterility,* 103, 6, 2015, 1418-1422.

[93] The paper is not referring to sex selection aimed to prevent the transmission of a particular disease, a possibility admitted in most national laws and in Article 14 of the Convention for the Protection of Human Rights and Biomedicine (Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine, https://rm.coe.int/168007cf98, last visited 20/04/2020).

[94] The same ban would apply to specific communities (Indian, Chinese) in Western countries. There was a study performed in Asian communities (from India, China, and South Korea) in the US that found that when these families had a girl as their first child the chance that the second child would be a boy was much higher than in other communities, suggesting that some kind of medical intervention was used to select a baby boy. Cf. D. FOX, *Interest Creep*, in *The George Washington Law Review*, 82, 2, 2014, 330.

that characteristic? Here we are faced with a dilemma. It can be said that despite the importance that feature has for a specific person, it is objectively irrelevant; therefore, there is no point in selecting it. But if so, there is no reason to forbid parents from choosing it. That is, if the desired feature does not influence the child's existence or its future possibilities in life, the State has no reason to intrude in this kind of parental decision. On the other hand, it might be the case that the feature is actually relevant for the child's development and future, and if so, parents should have something to say about it. Quoting John Harris, «if they are not important, why not let people choose? And if they are important, can it be right to leave such important matters to chance? ».[95]

6. The admissibility of some NHRRD

6.1. Reasons leading to the admissibility of NHRRD

Based on the arguments advanced in the previously section, it is difficult to sustain the complete legal ban of NHRRD.

The eventual admission of NHRRD is not based on the right to reproduce. As stated supra, the scope of protection of this right does not include the right to choose the type of child one may have, but simply the right to have a child free of serious illnesses or malformations. Accordingly, in view of reproductive rights, the only characteristics parents should be entitled to select are those that are health related. It is not possible to provide an exact definition of "health", but it should at least include the avoidance of serious and untreatable medical conditions.

There are three reasons for allowing NHRRD. First, it is not always possible to clearly distinguish the features related to health from all the others. Secondly, assessing the value of these features is quite complex. Some of them are as relevant to parents as the health condition of their offspring. Thirdly, there are no sound reason to forbid NHRRD. As demonstrated *supra*, the arguments usually invoked against NHRRD all fall short on their intent. In the legal world the rule is not that a conduct is forbidden unless there are reasons to allow it, but rather that the conduct is admissible unless there are reasons to forbid it.

6.2. Features to be selected

In theory, parents can select non-health related features objectively harmful to the future person, such as violent behaviours or a low level of intelligence. However, the selection of characteristics that would deprive the child of opportunities in his/her life and reduce his/her wellbeing should not be allowed.

Most of the features referred in this paper will indeed improve children's quality of life. A reasonably good state of health is not the only characteristic able to provide people a good standard of living. This was recognized by the Nuffield Council on Bioethics on its report on genome editing from 2018, which systematically invokes the notion of welfare (for existing and future generations):

[95] John Harris manifests his concordance in relation to the selection of those attributes because he does not qualify this practice as discriminatory. Cf. J. HARRIS, *Rights and Reproductive Choice*, cit.

ISSN 2284-4503

> In our view, what is morally important about human beings is not dependent on the possession of a particular set of genomic variations: we find the concept of 'the human genome' to lack coherence in any case. We conclude that so long as heritable genome editing interventions are consistent with the welfare of the future person and with social justice and solidarity, they do not contravene any categorical moral prohibition.[96]

The concept of "welfare" clearly surpasses the mere notion of "health". It can even embrace features traditionally considered enhancement, such as intelligence.[97]

6.3. Offspring selection and the destruction of human life

The procedure use in the NHRRD is of utmost importance because it can change the ethical and legal assessment of the decision.

Pre-conception scientific procedures are less problematic because they do not have to overcome constraints based on the protection of the unborn. When genetic selection takes place before conception (in other words, in gametes),[98] the unborn's protection cannot be invoked to forbid it, given that at this time a "person" does not yet exist. As for gametes, they are not human life, and therefore the protection afforded to human beings does not apply to them.[99] Thus, at this moment the range of parental choices is wider.

More problematic is when in vitro embryos are already created. At this stage embryos might have to be destroyed for not presenting the desired features. Due to the potential destruction of human life some jurisdictions have (almost) completely banned any kind of embryonic selection. For instance, the German Embryo Protection Law of 1991[100] forbids sex selection, except to avoid the transmission of a disease (par 3), and in its original version it also banned PGD. However, after a 2011 reform, Germany came to allow it, although with restrictions and only regarding severe medical conditions (par 3a).[101]

In the past couple of years, several reforms have emerged in Europe to relax the rules on PGD and allow embryonic selection under certain conditions.[102] However, PGD is never allowed for non-health

[96] THE NUFFIELD COUNCIL ON BIOETHICS, Genome Editing and Human Reproduction: Social and Ethical Issues, 2018, 158, http:// nuffieldbioethics. org/ project/ genome- editing- humanreproduction (last visited 23/04/2020).

[97] C. GYNGELL, H. BOWMAN-SMART, J. SAVULESCU, *Moral Reasons to Edit the Human Genome: Picking Up from the Nuffield Report*, in *Journal of Medical Ethics,* 45, 2019, 518.

[98] By selecting donors with certain characteristics parents can indirectly select the features of the child (see V.L. RAPOSO, *Wrongful Genetic Connection,* cit., and the case law therein referred). This is, nonetheless, a genetic lottery, as the child might not inherit those characteristics.

[99] M.T. BROWN, *The Potential of the Human Embryo,* in *Journal of Medicine and Philosophy*, 32, 6, 2007, 599-603.

[100] Act on the Protection of Embryos (Embryonenschutzgesetz - ESchG) of 13 December 1990, Bundesgesetzblatt 1990 Part I pp. 2746-2748, amended by Article 1 of the Act of 21 November 2011 (Bundesgesetzblatt 2011 Part I p. 2228, https://www.drze.de/in-focus/stem-cell-research/modules/the-german-embryo-protection-act?set_language=en (last visited 06/04/2020).

[101] B.B. VON WÜLFINGEN, *Contested Change: How Germany Came to Allow PGD*, in *Reproductive BioMedicine and Society Online*, 3, 2016, 60-67.

[102] Bayefsky describes the reforms in Italy, Switzerland, France and the United Kingdom in M.J. BAYEFSKY, *op. cit.*

related features. For instance, both the Portuguese[103] and Spanish[104] laws on assisted reproductive techniques allow the use of PGD, but only to detect health conditions.[105] This is essentially the same rule stated in Article 12 of the Convention on Human Rights and Biomedicine.

The most problematic scenario concerns uterine embryos and foetuses, because they are a form of human life (unlike gametes) in a more developed stage of development (when compared with in vitro embryos), and thus they require stronger protection.[106] This reasoning is based on the theory of gradual protection of the unborn,[107] generally accepted by several courts in Europe. See in particular the two decisions on abortion from the German Constitutional Court (BVerfGE) at the end of last century: BverfGE, 39, 1 (1975) and BVerfGE 88, 203 (1993). Generally, abortion based on the unborn's features is legally restricted to its therapeutic aspects. Thus, the only available option for NHRRC is abortion by request. In the context of a legal system wherein abortion can be freely performed in the first weeks of gestation, even on a healthy foetus, there is no plausible ethical and legal justification, nor any effective mechanism, to forbid an abortion based on non-health related features. If the unborn does not have the parents' desired features,[108] no matter how futile those features are, abortion (by request) is still allowed,[109] because the idea underpinning abortion by request is that no justification is required[110] (and it is fair to assume that few parents would publicly disclose that the real reason to abort was a non-health related feature, such as gender). That said, there is a distinction between abortion and other forms of offspring selection.[111] Abortion involves the destruction of human life at a later stage of development, and so it faces more legal and ethical restrictions than when in vitro embryos are involved or, *a fortiori*, gametes alone, which are not even a form of human life.

Gene editing is another mechanism for NHHRD. It can be used at any stage of development: prior to or after birth, and even prior to conception (on gametes). From the perspective of human life protection, gene editing raises fewer issues because gene editing does not involve the destruction of human life (unlike it happens with abortion and with PGD),[112] but rather the modification of an existing

[103] Articles 7, 28 and 29 of Law n. 32/2006, from 26 July, http://www.pgdlisboa.pt/leis/lei_mostra_articulado.php?nid=903&tabela=leis (last visited 22/03/2020).

[104] Article 12 of Law 14/2006, from 26 May, https://www.boe.es/buscar/act.php?id=BOE-A-2006-9292 (last visited 02/06/2020).

[105] For a discussion of the use of PGD to screen non-health related features, see B. WILLIAMS, *op. cit.,* 1311 ff.

[106] M.T. BROWN, *op. cit.,* 603-605.

[107] C.M. ROMEO CASABONA, *El Derecho a la Vida: Aspectos Constitucionales de las Nuevas Biotecnologías, in Tribunal Constitucional (ed.), Actas de las VIII Jornadas de la Asociación de Letrados del Tribunal Constitucional*, Madrid, 2003, 40 ff.

[108] "The question is whether something which is not positively in a child's interest can be tolerated or permitted if it is not positively against the child's interests" (R. ASHCROFT, *Bach to the Future: Response To: Extending Preimplantation Genetic Diagnosis: Medical and Non-Medical Uses*, in *Journal of Medical Ethics*, 29, 4, 2003, 217-219.

[109] However, some US states have banned abortion based on gender, race or the results of pre-natal diagnosis.

[110] Studies have shown the perils of "free" abortion: in a study quoted by Lori Andrews (L. ANDREWS, *Brave New Babies*, 2007, http://www.pbs.org/independentlens/frozenangels/babies.html, last visited 12/06/2020), 12% of parents declared that they would abort a foetus if they knew it had a propensity for being obese.

[111] This difference is pointed out in L.A. VACCO, *op. cit*., 1219.

[112] Pointing out the differences between gene editing and PGD, and favouring gene editing in his analysis, I. DE MIGUEL BERIAIN, *Is the 'Serious' Factor in Germline Modification Really Relevant? A Response to Kleiderman,*

genetic code. These notes make this method very appealing (but only when it reaches the required levels of safety and efficiency). However, just like the previous procedures, in the European scenario gene editing is also restricted to health-related features.[113]

To summarize, apart from cases of severe or incurable illness or malformation (i.e., health-related causes), the selection of all remaining features must be completed before conception (for instance, by means of gamete selection) and not after, to avoid the destruction of embryos and foetuses just because they lack the desired features.[114] To destroy an in vitro embryo carrying a severe medical condition or, *a maiori, ad minus*, to abort a foetus in that situation, is not comparable to the destruction/abortion of embryos and foetuses that are healthy but simply lack a preferred gender or eye colour.[115] The exception is gene editing because this procedure does not involve the destruction of human life (i.e., embryos, foetuses and those who are already born), so, it could be used in later stages when its safety is demonstrated.

7. Final considerations

The paper has argued that despite the existing legal prohibition in several jurisdictions – namely in Europe, the geographical focus of the paper – there are no sound reasons to ban NHRRD. The arguments commonly invoked against the selection of non-health-related features have demonstrated their fallibility.

The paper is not sustaining that NHRRD do not raise any legal issues, nor is it advocating that the law should allow these choices without any restriction or condition. Instead, the paper simply states that the general understanding that NHRRD should be banned by law must be reassessed, because the arguments usually raised against them are insufficient.

This is not a new discussion. The determination of offspring traits has been an ancestral aspiration, but the most recent discoveries and scientific achievements regarding the human genome have prompted the rebirth of this aspiration. The new genetic and reproductive techniques have brought with them the promise of a seductive and fascinating future, even though (it must be recognized) it may be a potentially dangerous one.

In a sense, genetic selection become unavoidable because competition among the human species compels parents to look for the "best child" they can have. New scientific acquisitions have agitated our traditional convictions. We have tended to believe that genes are supposed to be established by nature. However, we are on the verge of replacing genetic lottery with deliberate parental choice. After all, is serendipity really more valuable than planning?

Ravitsky and Knoppers, in *Journal of Medical Ethics,* 46, 2020, 151-152; G. CAVALIERE, *Genome Editing and Assisted Reproduction: Curing Embryos, Society or Prospective Parents*?, in *Medicine, Health Care, and Philosophy,* 21, 2, 2018b, 215-225.

[113] *Vide* Article 13 of the Convention for the Protection of Human Rights and Dignity and Article 13 of Spanish Law n. 14/2006.

[114] This is under the assumption that the unborn is not a person but is, nonetheless, a form of human life.

[115] A similar argument is made by B.L. WILDER, *Assisted Reproduction Technology: Trends and Suggestions for the Developing Law,* in *Journal of the American Academy of Matrimonial Law*, 18, 2002, 204.

Whether we like it or not, the future of humanity is in our hands now. Rather than fearing genetics, we should embrace it. We can do better than chance.[116]

[116] J. SAVULESCU, *The Maverick: It's our Duty to Have Designer Babies*, in *Reader's Digest,* 2012.

Special issue

What is wrong in extinguishing a species? Charting the Ethical Challenges of using Gene-Drive Technologies to eradicate *A. gambiae* vector populations

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ABSTRACT: This article analyses three ethical arguments against the use of gene-drive technologies to control for, and possibly extinguish, a particular species of vector mosquitoes (Anopheles gambiae) causing the malaria infection. We conclude that none of these arguments is truly persuasive in the specific case and, therefore, that using gene-drive technologies to suppress or eradicate the population of Anopheles gambiae could be ethically justifiable provided certain cautions referring to ecological consequences, evolutionary effects and social engagement of local communities.

KEYWORDS: Gene-Drive; genome-editing; bioethics; CRISPR-Cas9; biodiversity

SUMMARY: 1. Introduction – 2. A steep price to pay: the global burden of mosquito-borne diseases – 3. Scaling the genomic revolution to ecosystems: the power and promises of gene-drive technologies – 4. Three ethical arguments to object the use of gene-drives to extinguish *Anopheles gambiae* – 4.1 First Argument: species have intrinsic and absolute moral value – 4.2 Second argument: the moral value of species depends on the moral value of their individual components – 4.3 Third argument: the diversity of species contributes to the extrinsic value of biodiversity – 5. Conclusion and additional cautions.

1. Introduction

In this article we analyze the bioethical implications of using gene-editing and gene-drive technologies to eradicate one species of vector mosquitoes, *Anopheles gambiae*, which is the primary vector of malaria. What follows is divided in four main sections. Section two introduces the burden of mosquito-borne diseases and the need for new vector control strategies. Section three, then, outlines the historical development of biotechnologies for vector controls, from early attempts to exploit selfish genetic elements via breeding programs to recent CRISPR gene-editing techniques. On this basis, section four introduces and discusses three possible arguments according to which extinguishing a species could be considered unethical because: (i) all species have intrinsic and absolute moral value; (ii) species have a moral value that depends on the intrinsic moral value of the individuals belonging to them; (iii) the diversity of species is important as it contributes to the extrinsic value of biodiversity. As we argue, provided certain conditions are met, none of these

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arguments are persuasive in opposing the use of gene-drive technologies to eradicate the *Anopheles gambiae,* a public health intervention that might save millions of human lives over the next years.

2. A steep price to pay: the global burden of mosquito-borne diseases

Due to the SARS-CoV-2 pandemic, a virus that amplified in bats and then transferred to humans through an as yet unidentified intermediate species, the issue of animal-borne diseases (zoonoses) has finally assumed a central role in the scientific and general debate on public health. However, only a relatively small number of species causes significant harms to humans today. Of these, most prominent are those that reduce agricultural output and that cause or transmit diseases.[1] In this respect, the burden of mosquito-borne diseases such as malaria has a prominent role. According to the World Health Organization (WHO), in 2019 over 400.000 persons have died of malaria, the 67% of which were children under the age of five.[2] Furthermore, malaria has other consequences for human health and wellbeing. In 2018 more than 11 million pregnant women have contracted malaria, causing the birth of 900.000 underweight children with higher risks of premature death and developing severe chronic conditions.[3] Of the 24 million of children who contracted the infection in 2018, it has been estimated that nearly 2 million have developed severe or moderate anaemia.[4] Aside from malaria, mosquitoes are also the primary vector of other zoonoses such as the yellow fewer, zika, chikungunya, and dengue.[5] None of these is deadlier than malaria, but their combined effect is estimated to be responsible for 725.000 human deaths a year.[6] It has been supposed that mosquitoes could have been the deadliest animal in human history.[7]

Furthermore, although the memory of the malarial areas in countries such as Italy is still alive, today mosquito-borne diseases disproportionally affect poor and vulnerable populations that already carry a significant burden of socio-economical and health inequalities. In 2018, "85% of global malaria deaths […] were concentrated in 20 countries in the WHO African Region and India; Nigeria accounted for almost 24% of all global malaria deaths, followed by the Democratic Republic of the Congo (11%)".[8] Unsurprisingly, reducing the burden of mosquito-borne diseases has long been

[1] See A. BURT, *Site-specific selfish genes as tools for the control and genetic engineering of natural populations,* in *Proc. Biol. Soc.,* 270, 1518, 2003, 921-928.

[2] World Health Organization, *World Malaria Report 2019*, https://www.who.int/publications-detail/world-malaria-report-2019 (last visited 28.01.21).

[3] *Ibidem.*

[4] *Ibidem.*

[5] On the current burden of dengue in Africa, see E. MANCINI, R.M. ZAGARELLA, *Modelli deliberativi per l'allocazione delle risorse in sanità: il caso della dengue in Tanzania*, in *Medicina e Morale*, 3, 2019, 313-335.

[6] https://www.who.int/southeastasia/news/opinion-editorials/detail/towards-a-mosquito-free-monsoon (last visited 28.01.21); see also Comitato Etico Fondazione Umberto Veronesi, *Gene-drive e responsabilità ecologica. Parere del Comitato Etico a favore della sperimentazione con popolazioni di zanzare geneticamente modificate,* cit.

[7] https://www.theguardian.com/global/2016/feb/10/should-we-wipe-mosquitoes-off-the-face-of-the-earth (last visited 28.01.21); see also A. ZIELINSKI, *The Ethical Risks of Engineering Mosquitoes into Extinction to Stop Zika*, in *Think Progress*, 2016, https://thinkprogress.org/the-ethical-risks-of-engineering-mosquitoes-into-extinction-to-stop-zika-88e45e538d25/ (last visited 28.01.21).

[8] World Health Organization, *op. cit.*

identified as a priority by many institutions and non-governmental organizations committed to reducing global health inequalities.

In general, the efforts to reduce the damages of mosquito-borne diseases have pursued one or more of the following strategies. One is to rely on public-health interventions, from educational programs to enhance health-literacy to the distribution of Insecticide-Treated bed Nets (ITNs). Multiple controlled studies have suggested that ITNs are highly cost-effective in reducing malaria infections and in saving human lives.[9] Despite these encouraging results, however, it is unlikely that this class of interventions may lift the entire burden of mosquito-borne diseases unless it is integrated with other strategies.

A second strategy, then, is to develop effective measures and therapies to prevent or directly treat mosquito-borne diseases. This requires significant investments and years of research. Furthermore, results are not guaranteed: at present there still exists no cure for mosquito-borne diseases such as zika and chikungunya. However, even when effective treatments are available, other factors may prevent or reduce their ability to control or end an epidemic. In this respect, the case of malaria is telling. Today, malaria can be cured if promptly diagnosed and treated.[10] However, in the regions where malaria is endemic diagnostic and treatment options are often lacking or inaccessible, further exacerbating inequalities in access to health. A pilot study is under way to test the first malaria vaccine (RTS,S/AS01, or RTS,S) in three African countries; it will be completed in 2023. In 2014 the RST,S has been tested in a Phase 3 clinical trial that demonstrated that "among children who received four doses, the vaccine prevented approximately 4 in 10 (39%) cases of malaria and 3 in 10 (29%) cases of severe malaria over a four-year period".[11] Due to its relative efficacy, the vaccine is considered a potentially important complementary tool for malaria control efforts rather than a candidate solution for the zoonosis.

Finally, a third strategy is to control for vector populations, mainly insects. Most diseases are transmitted only by a few species of mosquitoes and parasites: in the case of malaria, the main vector is *Anopheles gambiae,* while in the case of dengue, yellow fewer, zika, and chikungunya is *Aedes aegypti.*[12] Reducing or eradicating these vector populations would reduce or eliminate the burden of associated diseases.[13] So far, however, attempts to control vector mosquito populations

[9] https://www.givewell.org/international/technical/programs/insecticide-treated-nets (last visited 28.01.21).

[10] https://www.who.int/activities/treating-malaria (last visited 28.01.21).

[11] On the ongoing studies for the malaria vaccine see Center for Vaccine Innovation and Access, *The RTS,S malaria vaccine*, https://bit.ly/3rW99ug (last visited 28.01.21).

[12] To be more precise, "Malaria in humans results from infection by any of five species of Plasmodium: P. falciparum, P. vivax, P. ovale, P. malariae, and P. knowlesi. These are transmitted to humans by approximately 50 species of mosquitoes, all belonging to the genus Anopheles. In sub-Saharan Africa, the vast majority of deaths are caused by P. falciparum transmitted by An. gambiae and the closely related An. arabiensis"; J.M. MARSHALL, C.E. TAYLOR, *Malaria Control with Transgenic Mosquitoe*s, in *PLoS Med*, 6, 2, e1000020, 0164.

[13] According to the WHO, vector control interventions "have one of the highest returns on investment in public health. Effective vector control programmes that reduce disease can advance human and economic development. Aside from direct health benefits, reductions in vector-borne diseases will enable greater productivity and growth, reduce household poverty, increase equity and women's empowerment, and strengthen health systems". World Health Organization, *Global Vector Control Response 2017-2030,* 2017, 15.

have achieved mixed results.[14] In part, this has been due to the limited efficacy of traditional methods. Pesticides, for instance, are relatively effective in killing mosquitoes. Yet, over time, mosquitoes tend to evolve forms of pesticide resistance. Furthermore, the use of pesticides has long-term negative impacts on human health and ecosystems[15]. Other traditional methods (i.e. fumigation of affected areas; elimination of breeding habitats, etc.) yield similar results: some are relatively effective, but alone they can hardly be expected to eradicate mosquito-borne diseases[16].

For these reasons, in the last decades, the search for alternative vector control approaches has increasingly turned to the life sciences, and in particular to molecular biology and genetic engineering.

3. Scaling the genomic revolution to ecosystems: the power and promises of gene-drive technologies

The development of biotechnologies to control vector populations has long been in the making. A first turning point has occurred by the mid of the 20th century, when scientists began to study selfish genetic elements able to circumvent the ratios of Mendelian inheritance and force the dynamics of gene frequency change in populations. In 1960 Craig and colleagues proposed a breeding program to favor a biased inheritance of a "male-producing factor" naturally occurring in *Aedes aegypti.*[17] When male mosquitoes carrying this factor breed, the resulting offspring is predominantly male. Thus, by releasing in the environment mosquitoes carrying this selfish element, it is theoretically possible to alter the normal sex-ration and to lower the population of females (the agents of zoonosis) below the frequency required for effective disease transmission. This proposal has been followed by other studies suggesting the use of selfish genetic elements to reduce vector populations, especially in invertebrate populations with short generations, and by the first mathematical model demonstrating how a desirable genetic element may spread to reach *fixation* in a target population.[18] Yet, at the time scientists lacked the molecular tools to engineer desirable genetic elements and associate them to biased mechanisms of genetic inheritance.

Then, after thirty years of research in molecular biology, in 1992 Kidwell and Ribeiro proposed the use of transposable elements (TEs) as a mechanism to drive an engineered gene in a mosquito

[14] On the different strategies currently used to fight malaria see ; see also The National Academies of Science, Engineering, and Medicine, *Gene Drives on the Horizon. Advancing Science, Navigating Uncertainty, and Aligning Research with Public Values*, Washington, 2016.

[15] "Synthetic pyrethroids and organophosphates, the two most commonly used sprayed pesticides, have been linked to multiple medical problems, including birth defects, cancer, and chronic dermatologic and respiratory health problems. […]. In addition to individual health risks, there is concern that insecticide spraying may have major unintended environmental impacts, including encouraging mosquito resistance and harming other insect species"; L. GREISMAN, B. KOENIG, M. BARRY, *Control of Mosquito-Borne Illnesses: A Challenge to Public Health Ethics*, in A.C. MASTROIANNI, J. P. KAHN, N.E. KASS. (eds.), *The Oxford Handbook of Public Health Ethics*, https://bit.ly/2RdKJQw, (last visited 28.01.21).

[16] On the limits of traditional approaches for vector control see World Health Organization, *op. cit.*

[17] G.B. CRAIG, W.A. HICKEY, R.C. VANDEHEY, *An inherited male-producing factor in Aedes aegypti*, in *Science*, 132, 3443, 1887-1889.

[18] The National Academies of Science, Engineering, and Medicine, *op. cit.*

population.[19] TEs are a special kind of selfish genetic element "able to spread quickly through a population due to their ability to replicate within a host genome and hence to be inherited more frequently in the offspring's genome".[20] Today we know that TEs are a fundamental source of genetic variation and have a prominent role in favoring genome plasticity. They not only produce deleterious mutations, but can promote the adaptability of species. For example, the genome of invasive species, including mosquitoes such as *Aedes albopictus*, are rich in TEs. Despite the initial excitement about TEs, the first studies evidenced some of their structural limitations, such as their low activity in vector mosquitoes, low replication rate, and vulnerability to losing internal sequences during replication.[21] To solve these issues, in 2003 Burt suggested the use of the homing endonuclease gene (HE), a site-specific selfish gene, to drive a specific genetic element in a population. HE was then at the centre of researches as possible basis for targeted gene therapy – an experimental approach in its early infancy. Burt proposed to extend these lines of research to vector control, arguing that HE had several advantages over previous candidates (such as TEs) due to its evolutionary stability, reversibility and applicability.[22]

These pioneering studies opened a new phase of research into so-called "gene-drives". In general, a gene drive can be defined as "a system of biased inheritance in which the ability of a genetic element to pass from a parent to its offspring through sexual reproduction is enhanced".[23] Under traditional Mendelian inheritance, the offspring has a 50% chance of inheriting a genetic element from a parent organism. Gene-drives are molecular accelerators that alter this ratio by inducing the preferential increase of a specific *genotype* – and, thus of the associated *phenotype* – "from one generation to the next, and potentially throughout the populations".[24] Also, these studies had the merit of calling for an open and public discussion over *the ethics* of gene-drives, and in particular "on the desirability of eradicating or genetically modifying particular species".[25]

These concerns acquired a new sense of urgency with the discovery of CRISPR (Clustered regularly-interspaced short palindromic repeats). CRISPR are segments of bacterial DNA that in conjunction with the guide protein Cas9 allow the editing of the genome of potentially any living organism. Gene-editing techniques based on CRISPR, in fact, allow for the insertion, deletion, copy and paste, or replacement of specific genes in a way that is sometimes orders of magnitude more precise, cheaper and simpler than previous techniques. Indeed, CRISPR gene-editing biotechnologies represent one of the major breakthroughs in the history of the life sciences due to their endless implications, in and

[19] Cfr. M.G. KIDWELL, J.M. RIBEIRO, *Can transposable elements be used to drive disease refractoriness genes into vector populations?*, In *Parasitology*. *Today*, 8, 10, 1992, 325-329.

[20] J.M. MARSHALL, C.E. TAYLOR, *op. cit*., 0165.

[21] A. BURT, *Site-specific selfish genes as tools for the control and genetic engineering of natural populations*, in *Proceedings Biological Science*, 270, 1518, 2003, 921-928.

[22] A. BURT, *op. cit.,* 922-23.

[23] The National Academies of Science, Engineering, and Medicine, *op. cit.*, 21.

[24] *Ibidem*.

[25] A. BURT, *op. cit,* 921.

beyond biology and biomedicine.[26] Being a more accessible technique, of course it also poses greater control problems.

Among their huge and multiple applications, CRISPR-based technologies provided a new and powerful method of engineering gene-drives as well. In 2015, different research groups were able to show that it was possible to use CRISPR to create specific gene-drives in yeast, fruit-flies and – most importantly – in vector mosquitoes.[27] Thus, only three years after its first demonstration as a gene-editing tools, CRISPR, paired with advanced knowledge about selfish genetic elements, "enabled a breakthrough in what scientists had been studying for more than 50 years".[28]

These studies also opened the possibility of creating different gene-drives to achieve diverse purposes. With respect to the use of gene-drives to control vector populations, two main approaches have been conceived. The first one aims at *population suppression*, that is, at spreading a genetic element that causes the number of individuals in a population to decrease, potentially driving it to extinction.[29] The second approach, instead, aims at *population replacement*, that is, at spreading a genetic element in a population that causes a population's genotype to change its frequency (e.g. spreading a gene that makes mosquitoes non-infectible by, or unable to transmit, dangerous pathogens such as *Plasmodium falciparum* causing malaria).[30]

Furthermore, these first studies have been crucial in highlighting and then overcoming various technical hurdles. One of the most pressing technical issue in the development of gene-drives for vector control is the emergence of counter-mutations that might reduce over time the driver capacity, producing resistance.[31] In this respect, an important step has been taken in a study in 2018 by Kirou et al. in which scientists used CRISPR to create a gene-drive targeting the *doublsex gene* (dsx).[32] This gene is crucial to ensure the reproduction of the species, it is strongly conserved by natural selection and therefore highly resistant to mutations. The studies have demonstrated that

[26] On the implications of gene-editing techniques for biomedicine, see Comitato Etico Fondazione Umberto Veronesi, *L'editing del genoma umano tra etica e democrazia*, in *The Future of Science and Ethics*, 3, 2018, 52-61.

[27] On the first studies to create gene-drives in mosquitoes with CRISPR see V.M., GANTZ, N. JASINSKIENE, O. TATARENKOVA, A. FAZEKAS, V.M. MACIAS, E. BIER, A.A. JAMES, *Highly efficient Cas9-mediated gene drive for population modification of the malaria vector mosquito* Anopheles stephensi, in *Proc. Natl. Acad. Sci.,* 112/E6736-E6743, 2015; and A. HAMMOND, R. GALIZI, K. KYROU, A. SIMONI, C. SINISCALCHI, D. KATSANOS, M. GRIBBLE, D. BAKER, E. MAROIS, S. RUSSELL, A. BURT, N. WINDBICHLER, A. CRISANTI, T. NOLAN, *A CRISPR-Cas9 gene drive system targeting female reproduction in the malaria mosquito vector* Anopheles gambiae, in *Nat. Biotechnol.*, 34, 1, 2016, 78-83.

[28] The National Academies of Science, Engineering, and Medicine, *op. cit.,* 13.

[29] *Ivi*, 16.

[30] Furthermore, it is also important to distinguish between *self-propagating* and *self-limiting gene-drives*; on this aspect see J. STEPHANIE JAMES, F.H. COLLINS, P.A. WELKHOFF, C. EMERSON, H. CHARLES, J. GODFRAY et al., *Pathway to Deployment of Gene Drive Mosquitoes as a Potential Biocontrol Tool for Elimination of Malaria in Sub-Saharan Africa: Recommendations of a Scientific Working Group*, in *Am. J. Trop. Med. Hyg.*, 98, 6, 2018, 1-49.

[31] On the technical difficulties of the first attempts to engineer robot gene-drives in mosquitoes see E CALLAWAY, *Gene drives thwarted by emergence of resistant organisms*, in *Nature*, 542, 7639, 2017.

[32] K. KYROU, A.M. HAMMOND, R. GALIZI, N. KRANJC, A. BURT, A.K. BEAGHTON, T. NOLAN, A. CRISANTI, *A CRISPR–Cas9 gene drive targeting doublesex causes complete population suppression in caged* Anopheles gambiae *mosquitoes*, in *Nature Biotechnology*, 36, 2018, 1062-1066.

the mutations created with CRISPR on *dsx* produced healthy males but sterile females in *Anopheles*. In just eleven generations the mutation has spread to the 100% of the individuals, leading to complete population collapse.[33] The speed of action is inversely proportional to the probability of developing resistance. Normally, starting from 600 mosquitoes after 11 generations, 20 million individuals are obtained; with the gene-drive on *dsx*, instead, after 11 generations the result was 0.[34] Furthermore, the resistant variants that have arisen in each generation have not blocked the spread of the driver. For this reason, the suppression gene-drive, whose killing action is faster, protects more from the risk of resistance. Following these successes, other researchers have begun to consolidate these results and planned the first experiments in controlled environments. Today, multiple gene-drive technologies based on CRISPR-Cas-9 and other genome-editing techniques have already been tested in controlled experiments. Some of these interventions will presumably soon be released in the environment, precipitating a host of pressing legal, political, regulatory, and ethical questions.

4. Three ethical arguments to object the use of gene-drives to eradicate Anopheles gambiae

CRISPR gene-editing biotechnologies coupled with gene-drive mechanisms provide powerful tools to control or suppress vector populations. However, the prospect of eradicating entire populations, varieties and species through these technologies is morally problematic and attracted a number of criticisms.[35] Although the development of gene-drives biotechnologies raises a host of complex ethical issues, in this section we limit our analysis to the potential moral objections of using gene-drive biotechnologies to extinguish one species of mosquitoes: *Anopheles gambiae*. More precisely, in this section we explore three possible ethical arguments against the eradication of *Anopheles*, arguing that they all fail to provide a convincing rationale to oppose this intervention.

4.1 First Argument: species have intrinsic and absolute moral value

According to Pugh, a first way to argue against the use of gene-drive technologies is to contend that each species has intrinsic and absolute moral value.[36] To exemplify this position, Pugh cites the entomologist R.L Metcalf, who, in objecting to the eradication of species as a method for pest control, noted that "species should be regarded as sacred and man indeed has no right to destroy them".[37] Pugh then also quotes M. Challenger – author of the best-selling book *On Extinction: How*

[33] *Ibidem*.

[34] *Ibidem.*

[35] For an overview of ethical challenges of gene-drives technologies see The National Academies of Science, Engineering, and Medicine, *op. cit.;* Comitato Etico Fondazione Umberto Veronesi, *Gene-drive e responsabilità ecologica. Parere del Comitato Etico a favore della sperimentazione con popolazioni di zanzare geneticamente modificate, cit.*; J. Stephanie James, F.H. Collins, P.A. Welkhoff, C. Emerson, H. Charles, J. Godfray et al., *op. cit.*

[36] See J. Pugh, *Driven to extinction? The ethics of eradicating mosquitoes with gene-drive technologies*, in *Journal of Medical ethics*, 42, i9, 2016, 578-581.

[37] See J. Perkins, *The Philosophical Foundations*, in J. Perkins (ed), *Insects, Experts, and the Insecticide Crisis: The Quest for New Pest Management,* 2012, 183-207; cited in J. Pugh, *op. cit.*

we became estranged from nature –, who observed (adopting the standard argument of the "slippery slope") that "I do think there's something more robust: the sanctity of life. If you start getting cavalier about the existence of a living being, if we start to think it's OK to eradicate something because it's a threat to us, we put other ideas about the sanctity of life in question".[38] Following a common categorization in bioethics, Pugh classifies both quotes under the category of the "sanctity of life" arguments against gene-drives. This label, however, is misleading. As shown by Jones, and contrary to a popular belief, the "sanctity of life" view is not tied to any religious tradition; it emerged in bioethics only in 1970s as a way of constructing a caricature of deontological positions over the permissibility of abortion and euthanasia.[39] Nevertheless, for the sake of the argument, let us depart from Pugh's reading as a way to introduce what we may label as the "expanded sanctity of life view". According to the general version of this view, extinguishing a species is always wrong, for *all biological life* has intrinsic and absolute moral worth, regardless of the species in question. Obviously, such a view is absurd. To hold that "all life forms have intrinsic and absolute moral value", in fact, would amount to hold a view that is factually incompatible with one's own life. We constantly kill other living organisms for our ends. The death of individuals and species by other individuals and species is at the basis of every ecological network and the evolution of every ecosystem. Of course, we are dramatically accelerating the extinction of other species, to the point that today we talk about the Sixth Mass Extinction of biodiversity, but this is not causally correlated with the planned and ethically motivated extinction of a single population or species of insects (see 4.3). We use plants for food and many other vital activities, and we kill bacteria with antibiotics.[40] Understood in this generalized way, the expanded view is "hopelessly inconsistent with everyday life". Hence, it must be rejected.

A more limited (and sensible) version of this view is to hold that only *species*, rather than all life forms have intrinsic and infinite moral value. According to this view, what is "sacred" is not life in general or individual organisms, but rather *species* in themselves.[41] On this view, killing one or more

[38] See https://www.theguardian.com/global/2016/feb/10/should-we-wipe-mosquitoes-off-the-face-of-the-earth (last visited 28.01.21); cited in J. PUGH, *op. cit.*

[39] D.A. JONES, *An unholy mess: why 'the sanctity of life principle' should be jettisoned*, in *The New Bioethics*, 22/3, 2016, 185-201. The tile of Jones's article is already explicit; in it he concludes: "The language of 'sanctity', in relation to the ethics of killing, emerged in a modern context to encourage in legal, ethical and theological discussion a false dichotomy between a supposedly religiously-based taboo (sanctity of life) and the making of prudential decisions about life sustaining treatment (quality of life). [T]o lend support that the phrase 'sanctity of life' serves only to perpetuate a discourse that is both confused and dangerous […]. Given that mistaken or contradictory conceptions of 'the sanctity of life' are ubiquitous, it is better to recognise that this is an unhelpful place to start"; see D.A. JONES, *op. cit.*

[40] See J. PUGH, *op. cit*. As P. Singer noted "People often say that life is sacred. They almost never mean what they say. They do not mean, as their words seem to imply, that life itself is sacred. If they did, killing a pig or pulling up a cabbage would be as abhorrent to them as the murder of a human being. When people say that life is sacred, it is human life they have in mind"; P. SINGER, *Practical ethics*, 1993, Cambridge, 83.

[41] Pugh suggests that it is in this sense that one should read Metcalf's remark that we must regard species *as if they were sacred.* A more sensible reading of this quote, however, is that it is not concerned with *moral value*, but with *respect* and *reverence*. On this aspect, Jones noted "Similarly the traditional prohibition on medical homicide does not imply that greater length of life is always the highest value, to be achieved at any cost and by any means. It is, rather, the claim that it is contrary to medical ethics intentionally to end the life of an

mosquitoes in our backyard could be morally permissible, while extinguishing *Anopheles gambiae* as a whole species would be not. Yet, also this view faces insurmountable theoretical issues. First, it is premised on the idea that a species can possess moral status as a collective entity regardless of the moral status of the individuals that belong to it. But the claim that "moral status" is an emergent property in the case of species is not self-evident and requires further explanation[42]. Second, and more importantly, the claim that all species are "sacred" – i.e. possess intrinsic and absolute moral value – is problematic for it entails that a species of bacteria, yeasts, mosquitoes or plants has *the same moral value* as *Homo sapiens*. Hence, to hold that all species have "intrinsic and absolute moral worth" entails that humanity has the same moral value as a species of yeasts, or plants. This is, at best, a highly counterintuitive conclusion, and which imply the somewhat controversial possibility that human beings can renounce any privileged point of view about themselves and their own evolutionary interests (the same paradox of those who would like to attribute rights, *human* rights, to plants and bacteria). In fact, sometimes eradicating another species may represent a desirable and justifiable aim. Consider, for instance, the case of the eradication of smallpox. Smallpox is fatal in nearly 30% of the cases and it has been estimated that it has killed 300 millions of human beings in the twentieth century alone. Smallpox was finally eradicated in 1980 following extensive vaccination campaigns. In this case, it is undeniable that eradicating the *Variola major* and *Variola minor* – the species of viruses causing smallpox – has been not only one of the greatest accomplishments of contemporary medicine but also a desirable and ethically justifiable outcome. Yet, if the adherent of the expanded sanctity of life view is understood to claim that "all living species are sacred", "then this vaccination programme would have amounted to a morally abhorrent form of 'speciecide'".[43] Again, a conclusion that would sound grossly exaggerated and counterintuitive to most.

Therefore, it seems that even under this second more charitable reading the expanded view of the sanctity of life is ultimately untenable. One cannot convincingly object to the use of gene-drive technologies to eradicate *Anopheles gambiae* on the grounds that either "all life" or "all species" are sacred – if by "sacred" one means that they possess *intrinsic* and *infinite* or absolute moral value. The consequence would be to make human life impossible, or to fall into the illusion of being able to renounce any form of anthropocentrism.

4.2 Second argument: the moral value of species depends on the moral value of their individual components

A second argument, then, is to contend that extinguishing a species may be morally wrong not because species have intrinsic moral value, but because *individuals* belonging to them have intrinsic moral value (or status). On this view, extinguishing *Homo sapiens* would be wrong simply because it would be wrong to kill billions of human beings who have moral value, and conversely, killing a single

innocent human being. Rather than the inviolability of life being an account of the value of length of life, it is simply a recognition of the disrespect that is shown to life when it deliberately destroyed"; see D.A. JONES, *op. cit.*

[42] Scholars like Russow, for instance, have provided convincing arguments to oppose the view that "obligations to a species might arise out of certain putative rights or interests of a species"; cfr J. PUGH, *op. cit.*

[43] J. PUGH, *op. cit.*

human being is like killing humanity itself. Peter Singer, in his classic *Practical Ethics*, defends a similar position as he criticizes the view of deep ecologists such as Bill Devall and George Session who attribute intrinsic and infinite moral value to entities such as *ecosystems* and *species*. As Singer concluded, it is better to "confine ourselves to arguments based on the interests of sentient creatures, present and future, human and non-human", for arguments centred on "holistic entities" are either metaphorical or fall into the same objections of the positions based on the sanctity/reverence of life.[44] Following these remarks, in this section we discuss some implications of Singer's utilitarian view in relation to the ethics of eradicating the species *Anopheles* via gene-drive technologies.

First of all, according to Singer, only *sentient* non-human animals have moral value, as they are the only creatures capable of having "morally relevant interests". More precisely, on Singer's view, non-human animals possess morally relevant interests only if they possess, at least, the minimal capacities "to suffer or experience enjoyment or happiness". Indeed, humans – as well as other primates, pigs, birds, or fishes – normally possess such capacities; therefore, they all have "morally relevant interests" in not being harmed or killed. In other cases, however, the issue of whether non-human animals possess or lack the minimal capacities to be sentient, and therefore to have morally relevant interests, is less clear-cut. In this paper, we focus our bioethical analysis only on invertebrates, namely mosquitos, and not on vertebrates and mammals such as invasive rodents, for which eradication interventions from islands (i.e. Floreana, Galápagos)[45] are being planned. Thus, our question is targeted: do insects and mosquitoes have some degree of morally relevant interests in Singer's minimal sense? Here there are two possibilities. One is that mosquitoes are not sentient, and therefore they do not have *any* moral status. In this case, one could simply not rely on this argument to object to the use of gene-drive technologies to eradicate *Anopheles.* The other possibility is that mosquitoes have some of these minimal capacities, and therefore killing them would be *prima facie* wrong, as it would be to eradicate one of their species. Which of these alternatives is correct?

Answering this question is less straightforward than it might appear at first. Specifying and assessing the criteria for which non-human animals possess or lacks moral status is, in fact, a notoriously controversial task.[46] Furthermore, there exists an ongoing debate in entomology over whether, and

[44] "There is, of course, a real philosophical question about whether a species or an ecosystem can be considered as the sort of individual that can have interests, or a 'self' to be realised […] For it is necessary, not merely that trees, species, and ecosystems can properly be said to have interests, but that they have morally significant interests. If they are to be regarded as 'selves' it will need to be shown that the survival or realisation of that kind of self has moral value, independently of the value it has because of its importance in sustaining conscious life […], In this respect trees, ecosystems, and species are more like rocks than they are like sentient beings; so the divide between sentient and non-sentient creatures is to that extent a firmer basis for a morally important boundary than the divide between living and non- living things, or between holistic entities and any other entities that we might not regard as holistic"; P. SINGER, *op. cit*, 82-83.

[45] K.J. CAMPBELL, J.R. SAAH , P.R. BROWN , J. GODWIN , F. GOULD et al., *A potential new tool for the toolbox: assessing gene drives for eradicating invasive rodent populations*, in C.R. VEITCH, M.N. CLOUT, A.R. MARTIN, J.C. RUSSELL AND C.J. WEST (eds.), *Island invasives: scaling up to meet the challenge*, 2019, 6-14.

[46] Today there exists a plurality of accounts identifying a wide range of capacities and properties that might provide the basis to attribute "moral status" or "standing" to non-human animals – such as sentience,

to what extent, non-human animals like mosquitoes, fruit flies and similarly complex organisms have or not the minimal capacities to feel pain, suffer, and in general to experience and process nociceptive stimuli.[47] Interestingly, for our present purpose, we can largely remain agnostic over these intricate issues. The reason is that gene-drive technologies operate in a fundamentally different way than other vector control strategies like pesticides. To see how this aspect makes a practical difference in the context of the present analysis, let us assume, for the sake of the argument, that mosquitoes have some minimal moral interests. This would entail, at least, that mosquitoes have the capacities to experience pain and suffering and therefore an interest in not being harmed or killed.

Interestingly, even if this were the case, a utilitarian adopting Singer's view would still lack a convincing rationale to oppose the use of most gene-drive technologies to eradicate the *Anopheles*. Gene-drive interventions, in fact, do not necessarily cause mosquitoes to experience suffering or die prematurely.[48] Rather, a species that has been genetically modified to spread a gene-drive like the one tested by Kirou et al. (2018) – which skews the sex-ratio by causing all female mosquitoes to be sterile – would simply be phased out over time. If these techniques were implemented successfully, "individual mosquitoes would live and die in the same way that they would have done in the absence of the intervention; they will just fail to reproduce", without being aware of it.[49] Therefore, even if we assume that mosquitoes have minimal moral interests, this would still not provide a convincing reason to object to the use of gene-drive technologies for population suppression – at least, from the point of view of Singer's utilitarian view.[50] Unless we think mosquitoes can suffer from being sterilized, which seems frankly absurd.

Moreover, adopting Singer's view would lead to a second, and decisive, series of considerations in favor of gene-drive interventions. Singer is one of the most influential proponents and advocates of the principle of the "equal consideration of interests" and, consequently, of *anti-speciesism.*[51] According to this principle, one should equally consider all affected interests when calculating the

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rationality, moral agency, dignity, and/or the capacities to feel pain, pleasure and emotions; for an overview see L. GRUEN, *The Moral Status of Animals*, in E.N. ZALTA (ed.), *The Stanford Encyclopedia of Philosophy,* (Fall 2017 Edition), https://plato.stanford.edu/archives/fall2017/entries/moral-animal (last visited 28.01.21).

[47] See, for instance, M.K. THANG, Q. WANG, J. MANION, L.J. OYSTON, M. LAU et al., *Nerve injury drives a heightened state of vigilance and neuropathic sensitization in Drosophila,* in *Science Advances*, 5, 7, 2019, eaaw4099.

[48] There are, however, notable exceptions. For instance, in 2021 750 millions of transgenic mosquitoes will be released in Floria in an effort to reduce the vector populations of Zika. These transgenic mosquitoes have been created by a US operated company Oxitec. The specific genetic modification developed by Oxitec, however, does reduce the lifespan of male mosquitoes; see D.O CARVALHO, A.R. MCKEMEY, L. GARZIERA, R. LACROIX, C.A. DONNELLY, L. ALPHEY et al., *Suppression of a Field Population of Aedes aegypti in Brazil by Sustained Release of Transgenic Male Mosquitoes*, in *PLoS Neglected Tropical Diseases*, 9, 7, 2015, e0003864.

[49] J. PUGH, *op. cit.*

[50] It can, however, provides a good argument to chose one gene-drive intervention over others. In fact, other things beings equal, recognizing that mosquitoes have morally relevant interests would provide a powerful argument in favor of developing gene-drive technologies aimed at replacing their population rather than suppressing it.

[51] According to Singer, one should equally consider all affected interests when calculating the rightness of an action – regardless of other factors such as the sex or *species* of the subjects in question; see P. SINGER, *op. cit*, ch. 1-2.

rightness or wrongness of an action, regardless of other factors such as one's skin colour, sex, or *species*. Endorsing this principle, however, does not entail that all interests are equal, but only that they ought to be equally considered. "When we come to consider the value of life – Singer writes –, we cannot say quite so confidently that a life is a life, and equally valuable, whether it is a human life or an animal life. It would not be speciesist to hold that the life of a self-aware being, capable of abstract thought, of planning for the future, of complex acts of communication, and so on, is more valuable than the life of a being without these capacities".[52] One can thus be antispeciesist and recognizes that, other things being equal, the moral interests of humans weigh far more than those attributable to non-human animals lacking complex cognitive capacities, such as mosquitoes.

With respect to the ethics of gene-drive technologies, this implies that, in any case, the interests that might be attributed to *Anopheles* ought to be weighed against the interests of millions of human beings that will suffer and die, and be worse off in case effective gene-drive technologies are withhold. While it is difficult to provide an exact estimation in such cases, with respect to vector control for malaria is hard not to conclude that the moral interests – i.e. the capacity to suffer, think, plan for the future, and flourish over time – of humans are almost incommensurable with respect to those of mosquitoes whose lifespan is of maximum two weeks. Given the significant burden of disease of malaria, and the difference between the moral interests of human and mosquitoes, other things being equal, if we adopt Singer's utilitarian view the eradication of *Anopheles* appears not only *justifiable*, but also *desirable* and ethically motivated. If anything, the results of such utilitarian calculations would provide a compelling ethical rationale to develop and release gene-edited mosquitoes (together with all other preventive actions/interventions against health inequalities) and save human lives.

Thus, also this second utilitarian argument fails to provide a persuasive basis to argue against the eradication of *Anopheles* via gene-drive technologies. Even if one grants to mosquitoes some kind of minimally relevant moral interests – a claim that is very dubious –, given the principle of the equal consideration of interests one would still retain good reasons to promote, rather than withhold, the use of such biotechnologies.

A possible objection to this conclusion is that it is premised on the view that what matters are only the interests of presently living mosquitoes. What about the interests of the beings that will never be born because of the gene-drive technologies? This objection introduces a variant of the second argument for which species have a moral status that depends both on the moral status of presently and future living organisms belonging to them. This line of reasoning leads to intricate moral issues, which, again, we can fortunately sidestep in analyzing the ethics of gene-drive technologies for the case at hand given the incommensurable different weights between the moral interests of present and future humans and the presumed moral interests of present and future mosquitoes. Comparatively, from a utilitarian and pragmatic point of view, this problem would have been much more pressing if the non-human animals in question were clearly sentient (e.g. primates) and the number of human lives saved and/or of human suffering spared would have been not as significant as in the case of malaria.

[52] *Ivi*, 61.

4.3 Third argument: the diversity of species contributes to the extrinsic value of biodiversity

A third and possible argument against the use of gene-drive technologies to eradicate *Anopheles,* then, is that the diversity of the species contributes to the extrinsic value of *biodiversity*. Today, preserving biodiversity is considered an important value in environmental ethics as well as in biolaw. In general, the term "biodiversity" refers to the variety of all lifeforms.[53] Biodiversity may be defined in different ways, from molecular to ecological levels, but it is commonly understood as encompassing at least three dimensions: the diversity between all species, the genetic diversity within each species, and the diversity within and between ecosystems.[54] Currently, biodiversity is under lethal threat. Because of human actions (deforestation, invasive species, demographic growth, pollutions, over-hunting and over-fishing, climate change), species have been disappearing at 50-100 times the natural rate, and this loss is predicted to rise dramatically.[55] The loss of biodiversity provides an additional reason to evaluate with caution any intervention aimed at extinguishing a species. If we agree that to extinguish other lifeforms is unethical as it compromises biodiversity, then we would have a legitimate objection to the use of gene-drive technologies to suppress *any* species, including *Anopheles*.

On this basis, in this section we discuss a specific argument for which reducing biodiversity is morally wrong as it may jeopardize the survival and wellbeing of humanity, which depend on biodiversity and ecosystem services. The idea that biodiversity – and nature in general – must be preserved primarily for the sake of humanity is, of course, not new.[56] The 1992 *Convention on Biological Diversity*, for instance, affirms that all contracting parties recognize the "intrinsic value of biological diversity and of the ecological, genetic, social, economic, scientific, educational, cultural, recreational and aesthetic values of biological diversity and its components".[57] This passage seems to attribute both an intrinsic as well as an extrinsic value to biodiversity. However, in this section we shall limit our discussion only to the latter aspect.[58] As the Secretariat of the Convention explains in another document, "[p]rotecting biodiversity is in our self-interest. Biological resources are the pillars upon

[53] For an overview of the history of the term "biodiversity", and of the conceptual issues arising from the various attempts at defying it in contemporary philosophy of biology and environmental ethics see D. FAITH, *"Biodiversity"*, in E.N. ZALTA (ed.), *The Stanford Encyclopedia of Philosophy* (Fall 2019 Edition), https://stanford.io/39NWALw , (last visited 28.01.21).

[54] Cfr. Secretariat of the Convention on Biological Diversity, *How the Convention on Biological Diversity promotes nature and human well-being*, 2000, https://www.cbd.int/convention/guide/?id=web (last visited 28.01.21).

[55] *Ibidem*. See also: T. PIEVANI, *Earth's Sixth Mass-Extinction Event*, in *Science Direct*, Elsevier – Online Reference Database: Earth Systems and Environmental Sciences, online 9, 2015, doi 10.1016/B978-0-12-409548-9.09216-2.

[56] See P. SINGER, *op. cit*, 265-268.

[57] United Nations, *Convention on Biological Diversity,* 1992 https://www.cbd.int/doc/legal/cbd-en.pdf (last visited 28.01.21). The *Convention* was opened for signature at the United Nations Conference on Environment and Development in Rio de Janeiro in June 1992 and has as today has been ratified by over 150 countries, providing and important reference for international laws over ecological matters.

[58] It is also worth noting that specifying in which sense "biodiversity" could or should be considered something intrinsically valuable from a moral point of view is not an easy task. On this issue, see, for example, D. FAITH, *op. cit*.

which we build civilizations. Nature's products support […] diverse industries [….]. The loss of biodiversity threatens our food supplies, opportunities for recreation and tourism, and sources of wood, medicines and energy". Because of the instrumental value of biodiversity, this document concludes that it is "unethical to drive other forms of life to extinction, and thereby deprive present and future generations of options for their survival and development". With respect to the ethics of gene-drive technologies, the relevant question to ask is, therefore, what impact the eradication of *Anopheles* could have on biodiversity and for present and future generations.

In answering this question two considerations are in order. First, from the point of view of the diversity between species, if these interventions were successful, then one species of mosquitoes – the *Anopheles* – would likely cease to exist. However, it should be noted that there are more than 3500 known species of mosquitoes, the *Anopheles* being just one of them. Eradicating the *Anopheles*, then, would not be tantamount to extinguish *all* mosquitoes, but only *one* of their many species. Moreover, according to ecological and evolutionary principles, the use of gene-drive technologies may also favor other species of non-zoonotic mosquitoes and insects living in the same areas – e.g., by reducing the current use of insecticides as anti-malaria strategies and/or by freeing new ecological niches.

Second, assessing the wider implications of an intervention that takes place at the level of an entire species and ecosystem is a complex and difficult task. Indeed, "[r]eleasing gene drives into the environment means that complex molecular systems will be introduced into complex ecological systems, setting off a cascade of eco-evolutionary dynamics. Key considerations include fitness, species dispersal, gene flow, ecosystem dynamics, and evolution"[59]. What kind of effect should be expected on the ecosystem if we decide to release gene-edited *Anopheles* mosquitoes?

In the last years, a plethora of rigorous empirical studies have been conducted with the aim of answering this kind of questions and estimating the environmental implications of extinguishing *Anopheles g.* species. Significantly, a recent and comprehensive review of the existing literature has concluded: "*Anopheles gambiae* is a species of importance because of its role as a vector of malaria, not as a key component of ecosystem food webs. […] Adult *An. Gambiae* mosquitoes are a relatively low-value, low-volume and disaggregated resource and this is reflected in a lack of evidence for any tight links with predators […] This generalist predation is a known stable strategy in ecological theory and contributes to dynamic equilibria in predator and prey populations and in the ecosystem in general. Several competing mosquito species could increase if *An. gambiae* density is reduced in specific habitats. Many generalist predators of *An. gambiae* already prey on these species and would substitute them for *An. gambiae* if the latter were less abundant. In this sense, any positive effects of competitive release on abundances of other mosquito species have the potential to compensate for any reduction of *An. gambiae* biomass in a diet".[60]

Thus, according to available empirical evidence, it seems that the impacts of eradicating *Anopheles* on biodiversity is likely to be very low, for no other insectivorous species is entirely dependent on

[59] The National Academies of Science, Engineering, and Medicine, *op. cit*., 34.

[60] C.M. COLLINS, J.A. BONDS, M.M. QUINLAN, J.D. MUMFORD, *Effects of the removal or reduction in density of the malaria mosquito, Anopheles gambiae s.l., on interacting predators and competitors in local ecosystems*, in *Medical and Veterinary Entomology*, 33, 2019, 10.

ISSN 2284-4503

them for its own survival. Prudentially, considering the complexity of ecological systems, this prediction will have to be carefully verified first in controlled environments, and then through controlled release phases in localized environments, with stringent step-by-step controls. On the other hand, it is clear that the eradication of *Anopheles gambiae* could have a tremendous positive impact on present and future generation by reducing or eliminating the various health and economic burdens of malaria. In fact, we know that the poverty of the populations living in the areas with the highest biodiversity is itself a threat to biodiversity, which is seen as a subsistence resource for those who have nothing to make a living. Other things being equal, then, if the primary value to be considered is the good of present and future generations, then the burden of proof should arguably fall those willing to resist the use of gene-drive interventions rather than on those proposing their adoption.

5. Conclusions and additional cautions

Given the burden of mosquito-borne zoonoses – and especially of malaria – in this article we have argued that the use of gene-editing techniques to create gene-drives finalized at suppressing *Anopheles g.* population is, at least, *prima facie* ethically permissible. In particular, we have showed that the use of such interventions cannot be convincingly objected from the point of three possible arguments based, respectively, on the claim that: (i) all life or all species have intrinsic and absolute moral value; (ii) extinguishing a species is necessarily wrong if the individuals comprising it have some degree of morally relevant interests; and (iii) extinguishing a species is ethically wrong as it jeopardizes biodiversity and thus the present and future of humanity. If this is the case, the gene drive techniques with the purpose of replacement and not of suppression (so, without extinction) seem even more ethically admissible, with the release of mosquitos genetically modified in order to no longer be carriers of malaria (though, given the technical risk of resistance to be controlled).

However, it should be emphasized that our perspective in favor of the use of gene-drives to eradicate *Anopheles* should be assessed by taking into account at least three considerations. First, that the validity of our conclusions apply only insofar as these technologies are used to eradicate population of vector mosquitoes carrying severe and fatal diseases. If the same techniques were be used to create gene-drives in other species, for example in agriculture or for vertebrate species such as invasive rodents, then the same arguments would lead to different conclusions. For instance, if instead of the eradication of *Anopheles* the same biotechnologies would be used to create a gene-drive in a sentient mammal species, then both the second and the third argument would need to be re-evaluated, as such a species might have significantly different moral interests as well as a different impact on biodiversity and thereby on human survival and flourishing.

Second, gene-drive is a biotechnology able of generating hereditary effects and therefore has evolutionary implications. Although reverse gene-drives are being studied, such interventions are irreversible so far. Another reason for caution is that gene-drives producing homozygosis during meiosis and accelerating changes in gene frequencies until fixation tend to reduce genetic variability

in natural populations, a precious asset that we should alter only with adequate arguments. Therefore, an ethic of responsibility is necessary for gene-drive applications.[61]

Third, even in the specific case of assessing the ethical impact of vector control strategies, the use of gene-drive technologies raises many other issues that ought to be properly addressed. These include, among many other important concerns, issues such as: the ethics of carrying out responsible experimentations in confined and controlled environments, in order to test the techniques for unintended consequences in the targeted environments; the ethics of respecting current International norms in scientific research about biosecurity and responsible innovation; the proper engagement of the local populations that live in areas in which the gene-edited mosquitoes will be released, as well as of other potential stakeholders.[62] We need a democratic alliance between advanced science and biotechnologies, environmental protection, and local communities. We should always consider these promising and powerful biotechnologies a common good at the service of communities, well-being and peace. They must certainly not be intended as an alibi for not intervening also on the social and economic aspects that favor the spread and permanence of devastating zoonoses such as malaria. As we have explained elsewhere, the use of new and powerful biotechnologies requires not only ethical awareness, but also a comprehensive effort that ought to consider many other social, economic and ecological aspects.[63]

[61] C. EMERSON, S. JAMES, K. LITTLER, F. RANDAZZO, *Principles for gene drive research*, in *Science*, 358, 2017, 1135-1136.

[62] On the various technical and ethical challenges of experimenting on gene-drive in mosquitoes population see J. STEPHANIE JAMES, F.H. COLLINS, P.A. WELKHOFF, C. EMERSON, H. CHARLES, J. GODFRAY et al*., op. cit.*; on the broader ethical and social challenges of these biotechnologies beyond the experimental phase see, instead, The National Academies of Science, Engineering, and Medicine, *op. cit.*

[63] Comitato Etico Fondazione Umberto Veronesi, *Gene-drive e responsabilità ecologica. Parere del Comitato Etico a favore della sperimentazione con popolazioni di zanzare geneticamente modificate, cit.*

ISSN 2284-4503

About coevolution of humans and intelligent machines: Preliminary notes

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ABSTRACT: Cooperation is something worthy to be explored from a social, economic, biological and even genetic point of view. This paper deals with human cooperation and focuses specifically on how humans interact with intelligent machines, which are considered as entities that, along with others (humans and non-human animals), populate the same ecological niche. The discourse is based on two theoretical pillars: the hypothesis of self-domestication of humans and the niche construction theory. Then, the movement of intelligent machines from isolation to direct cooperation is shown as the factual technological change which raises the problem of how cooperation between humans and intelligent machines works and with what effects. A presentation of the two main visions about the future of human-machine relations is offered and the different possibilities of development of self-control between humans and intelligent machines are discussed. According to the Author, machines will not destroy humanity. Humans will co-evolve with the machines they create which they will control through social, ethical, and legal rules. In addition, humans, integrated with mechanical or electronic devices, will continue their evolution by developing their self-control as cyborgs. A final note is reserved for how our ecological niche is changing.

KEYWORDS: Human cooperation; intelligent machines; robot; ecological niche; technological change

SUMMARY: 1. The point – 2. Two theoretical pillars: self-domestication and niche construction – 3. Intelligent machines: from isolation to cooperation – 4. Intelligent machines, humans and augmented humans: whose self-control? – 4.1. Robots, autonomous systems, intelligent machines – 4.2. Two competing visions on humans and intelligent machines – 4.3. Intelligent machines and self-control – 4.4. Augmented human intelligence – 5. Coevolution of humans-machines and our ecological niche.

1. The point

This paper deals with human cooperation and focuses specifically on how humans interact with intelligent machines, which are considered as entities that, along with others (humans and non-human animals), populate the same ecological niche.

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ISSN 2284-4503

The idea of this topic came from some conversations I had with Monika Gruter Cheney and Oliver Goodenough, November 2019 in Stanford-Palo Alto. They were thinking about "cooperation" as the issue of the 2020 Gruter Institute annual meeting. The question that came to my mind was: if cooperation is something worthy to be explored from a social, economic, biological and even genetic point of view, why not to extend this exploration also to the cooperation between humans and intelligent machines?

Finally, the 2020 round of Gruter Institute annual meeting was (for the first time remotely) held in October 15-17 and focused on *The evolution of cooperation* and on how cooperation works:

> Darwin is often said to have left cooperation "unexplained." Subsequent evolutionary biologists such as W.D. Hamilton have spent decades seeking to understand it. It is particularly important for humans. Cooperative groups allow the division of labor, the specialization of abilities, and productive trade. […] When you apply this approach across entire countries or even the globe, the productive power of groups of humans is huge. And the advantages are not just on the production side. Sharing of resources across groups provides important insurance against short term failure by any given member.[1]

In the *Topic Précis* of the meeting the organizers outline specific aspects of cooperation, such as the case of some kind of insurance contracts in our contemporary markets, which are exactly scaled up applications of the cooperation and distribution of risks, and, also, the extraordinary power of human exchange and law, as one of the most important domains of institutional design and creation. The talk I gave on *Humans, cooperation and artificial entities* is the basis of this paper.

It seems to me that the cooperation among humans is an extremely interesting issue, something worthy to be explored in its various ramifications, e.g., under biological and even genetic point of view. Epigenetics offers an insight for such broadening of the scope of cooperation. Indeed, epigenetics implies a shift of focus from the individual genetic makeup to the environment and its retroaction on the human genetic dowry.

In this paper I try to go a little bit forward and laterally, and to outline a reply to the broader question of how we, human beings, change our ecological niche and how it retroacts on human beings. I say *forward* because the exploration seems to be worth to be extended beyond interactions among humans and to include also human-nonhuman animals interactions,[2] and *laterally* because the cooperation between humans and intelligent machines (namely humans-machines interactions) is, in my view, the challenging point of the next future. In this paper I'll focus on this lateral side of the issue, referring for the theme of relations between humans and non-human animals in ethical and legal studies to a previous research of mine.[3]

[1] Gruter Institute for Law and Behavioral Research, *Topic Précis, The Evolution of Cooperation, Virtual Squaw Valley Conference*, October 15-17 2020. Organizers: Monika Gruter Cheney, Oliver Goodenough, Andrew Torrance, Isabel Behncke.

[2] A session in the meeting was dedicated to non-human primates, with the participation of Isabel Behncke, Cognitive and Evolutionary Anthropologist at Oxford University.

[3] A. SANTOSUOSSO, *The human rights of nonhuman artificial entities: an oxymoron?*, in *Yearbook of Science and Ethics/Jahrbuch für Wissenschaft und Ethik*, 2015.

In paragraph 2., the two theoretical pillars on which the discourse is based are briefly presented, i.e. the hypothesis of self-domestication of humans and the niche construction theory; then, in par. 3., the movement of intelligent machines from isolation to direct cooperation is shown as the factual technological change which raises the problem of how cooperation between humans and intelligent machines works and with what effects; in par. 4., premised on a clarification of the terms used (robot, autonomous system and intelligent machine), the two main visions about the future of human-machine relations are presented and the different possibilities of development of self-control between humans and intelligent machines are discussed. In par. 5. a final note is reserved for how our ecological niche is changing.

2. Two theoretical pillars: self-domestication and niche construction

Cooperation between humans and intelligent machines can be properly framed by relying on two lines of research that exist in the literature and can be considered the premises of this paper: the self-domestication hypothesis and the niche construction theory.

According to the self-domestication hypothesis, humans have gone through a process of selection against aggression, a process that was self-induced. In his book on *The Goodness Paradox*, Richard W. Wrangham notes as some human extraordinary abilities of present times, such as heart surgery, space travel, and comic opera, all depend "from an evolutionary point of view […] on capacities for a quite exceptional ability to work together, including tolerance, trust, and understanding. Those are some of the qualities that cause our species to be thought of as exceptionally 'good'".[4]

The fact that man's relative docility and tolerance is a rare phenomenon in wild animals justifies the idea that man is a domesticated species. So, the question turns into "who could have domesticated us?" A possible reply is we might have self-domesticated, as bonobos did. Similar to humans, bonobos "show many of the features of a domesticated species" and, excluding humans having domesticated them, the conclusion might be "the process happened in nature" (thus, they "must have self-domesticated").

However, the question of *how* self-domestication happened stands and is still open, even though some suggestions seem to come from the "way that aggressive individuals are prevented from dominating others" or the creation of "despotic" hierarchical social institutions, which cannot be found in other species. In addition, the same *why* self-domestication happened is still open, as the parallel with bonobos evolution does not explain "why bonobo aggression was reduced" (*ivi,* 103) and why it was reduced in humans as well.

The paradox of virtue and violence, both so prominent in human life, is a topic of Wrangham reconstruction:

> We have evolved in both directions simultaneously. Both our tolerance and our violence are adaptive tendencies that have played vital roles in bringing us to our present state. The idea that human nature is

[4] R.W. WRANGHAM, *The Goodness Paradox*, 4.

> at the same time both virtuous and wicked is challenging, since presumably we would all wish for simplicity. (*ivi,* 12).[5]

According to Wrangham, Jean-Jacques Rousseau and Thomas Hobbes are classic icons for the alternatives. Rousseau has come to stand for humanity's being instinctively nice, Hobbes for humanity's being naturally wicked. However, "both positions have some merit. There is plenty of evidence that humans have innate tendencies for kindness, just as there is for our having spontaneously selfish feelings that can lead to aggression. No one has found a way to say that one kind of tendency is more biologically meaningful or evolutionarily influential than the other".[6]

As intriguing as it is, the idea of some sort of immutable balance between aggression and kindness in humans seems to be contradicted by the lowering of violent crimes around the world reported in criminology.[7] Of course, less violence does not equal to less evil behavior, but also the opposite is not true, as violence cannot be said to be morally insensitive.

In a recent study, Dor Shilton et al. criticize the concept of self-domestication and maintain "the social evolution of humans is better explained in terms of selection for pro-social motivation and self-control, which are guided by symbolic communication and representation rather than as a process of self-domestication".[8] In this view a special role was played by the emergence of mimetic communication, the beginnings of musical engagement, and mimesis-related cognition, and, in a second stage, the emergence of language and imagination, which facilitated emotional control and emotional plasticity.

The authors highlight the ambiguity of the theory of domestication, which was popular in the literature about social hierarchies in civility (more civilized humans who domesticated inferior humans) and later racist and eugenic political movements. This well-known problem (stressed also by Wrangham) is the result of the intrinsic ambiguity of the concept of domestication when used in explaining human evolutionary processes. At the end, the authors "do not find the notion of human self-domestication useful and believe that the partial analogy with domesticates focuses too much on the reduction of reactive aggression and too little on social organization".

Of course, I make no claim to resolve the open questions in each of these two theories and, therefore, refer to both in the later part of this paper.

The second pillar of this paper is the niche construction theory. According to this vision of evolution, organisms are not passive entities, malleable at will by selection. The metabolic and behavioral activities of biological populations change the ecological niches, thus influencing the environmental resources and the selective pressures that in turn retroact on organisms themselves. This

[5] "For centuries, people have simplified their understanding of a confusing world by adopting one or the other of these opposed views".

[6] R.W. WRANGHAM, *The Goodness Paradox*, 5-6. See also Id., *Two types of aggression in human evolution*, in *Proc. Natl. Acad. Sci.,* U.S.A, 115, 2018, 245–253. doi: 10.1073/pnas.1713611115.

[7] M.R. SANTOS, A. TESTA, *Homicide is declining around the world because we're getting old,* in *Quartz*, November 11, 2019, https://qz.com/1743595/why-global-homicide-rates-are-declining/#:~:text=A

[8] D. SHILTON, M. BRESKI, D. DOR, E. JABLONKA*, Human Social Evolution: Self-Domestication or Self-Control?,* in *Front. Psychol.*, 11:134, 2020, doi: 10.3389/fpsyg.2020.00134.

ISSN 2284-4503

phenomenon, called "niche construction", is essential in evolution. Organisms actively change their environment and the environments selectively change organisms.[9]
The idea at the basis of niche construction theory is interesting as it is able to encompass the relation humans-machines in the wider concept of human evolution simultaneously with the larger environment and also the products of human activity, from machines till to social institutions as laws and regulations.

3. Intelligent machines: from isolation to cooperation

The case of robots in industry is meaningful. In past decades robots were kept in isolation. Now the main concept is to boost a closer interaction and cooperation between human workers and robots. In this light, robots enable humans to enhance their performance.
In the last ten years, some important changes happened in the relationship between society and Artificial Intelligence, robotics and advanced technologies. If, for example, we think of robots in industry, up to a certain point the prevailing idea was that they were potentially dangerous entities to be kept in confined places and departments, so that they would not harm humans. Since then, the idea has changed and progressively they started to be accepted as entities facilitating human work and replacing mainly the simplest and most repetitive tasks. And, what is most interesting, they started being considered as entities with which you could work together side by side. This was the result of several factors such as the development in safety devices and measures, and also in Artificial Intelligence increasingly embedded in tools, machines and systems.
In this new environment the driving idea has become that of cooperation. Some have given this new horizon the name of intelligent automation continuum, others have spoken of a new "Age of With".[10] Where the idea of *with* is understood as the co-presence and interaction between different technologies, such as character recognition systems (ICR and OCR), natural language processing (NLP) and natural language generation (NLG), and more.[11]
It is against this current (and predictable) backdrop that the question of cooperation in human evolution must be posed today, and the question of what shape niche construction might take.
The development of powerful and intelligent machines impacts the ecological niche in several ways. One is the environmental one, considering for example the huge amount of electricity those

[9] T. PIEVANI, *How to Rethink Evolutionary Theory: A Plurality of Evolutionary Patterns,* in *Evol Biol,* 2015, DOI 10.1007/s11692-015-9338-3; K. LALAND, B. MATTHEWS, M.W. FELDMAN, *An introduction to niche construction theory,* in *Evol Ecol.*, 30, 2016, 191-202.
[10] On *intelligent automation continuum* see S. KAPOOR, *IDC Perspective: Six Capabilities from Leading RPA Service Providers That Advance Financial Services Institutions Toward Intelligent Automation*, 2020, in: https://www.capgemini.com/wp-content/uploads/2020/04/An-IDC-perspective-on-Intelligent-Process-Automation.pdf (visited November 20, 2020). On the *Age of With* see the 2019 Deloitte Report, significantly titled *Automation with intelligence*, https://bit.ly/3lJOY1h (visited November 20, 2020).
[11] S. OVERBY, *How Robotic Process Automation (RPA) and digital transformation work together*, August 11, 2020, https://enterprisersproject.com/article/2020/8/how-rpa-robotic-process-automation-and-digital-transformation-work (visited November 20, 2020).

technologies consume (such as for example mining in blockchain or 5G),[12] but the one I wish to focus on is the interaction between humans and intelligent machines, in the light of the theory of self-domestication or self-control, as determining forces in the construction of the ecological niche. Indeed, the above outlined scenario can be considered a description of our ecological niche.

At a first schematic level the dilemma can be formulated as follows: should we domesticate intelligent machines? Or will intelligent machines self-domesticate or develop their own self-control?

The first horn of the dilemma is consistent with the traditional reassuring idea that machines are a mere tool to be used at will by humans and underestimate how the presence of those intelligent machines retroactively affects the way humans think and act. The second horn of the dilemma takes for granted the development of higher forms of artificial intelligence that can have human-like evolutions.

The question requires more in-depth considerations, which are carried out in the next paragraph.

4. Intelligent machines, humans and augmented humans: whose self-control?

In this paragraph, premised on a clarification of the terms used, and presented the two main visions about the future of human-machine relations, the different possibilities of development of self-control between humans and intelligent machines are discussed.

4.1. Robots, autonomous systems, intelligent machines

A preliminary clarification is necessary about wording: robot, autonomous system, intelligent system/machine express different concepts and refer to different entities.

According to its etymology, *robot* refers to a mechanical entity (a machine) which does an even cognitively poor work instead of humans,[13] while *autonomous system* stresses the specific feature of a machine which is able to act (at a certain degree) without human input or with a non-direct intervention/suggestion by a human.[14] Finally, an *intelligent system/machine* is an entity which, having a variable balance between its mechanical components and software or AI components, is able to work intelligently, within the limits of and according to its AI components and devices.[15]

[12] Although it could be said that it is not enough to be a new selective pressure, rather having effects only on the social niche.

[13] The word "robot" is first known to be found in the sci-fi drama R.U.R. (1920), by Czech writer Karel Čapek, set in Rossum's Universal Robots (R.U.R.) industry, which used human replicants (called robots, in fact) as labor slaves for humanity. The Czech neologism "robot" is derived from the word "robota" meaning "work".

[14] G. SARTOR, A. OMICINI, *The autonomy of technological systems and respon-sibilities for their use*, in N. BHUTA, S. BECK, R. GEISS, C. KRESS, H.Y. LIU (eds.), *Autonomous Weapons Systems: Law, Ethics, Policy*, Cambridge, 2016, 39-74. See also, RockEU Robotics Coordination Action for Europe*, ELS issues in robotics and steps to consider them Part 2: Robotics and Regulations*. The deliverable was coauthored by B. BOTTALICO, A. SANTOSUOSSO, O. GOODENOUGH, R. DE BRUIN, C. HOLDER, C. GÔME, Y. DE FRANCE, C. NIEL-AUBIN, N. BENDER, C. LEROUX and delivered on June 2016, available at https://bit.ly/3eiZrxt.

[15] A. SANTOSUOSSO, B. BOTTALICO, *Autonomous Systems and the Law: Why Intelligence Matters*, in E. HILGENDORF, U. SEIDEL (eds.), *Robotics, Autonomics, and the Law: Legal issues arising from the Autonomics for Industry 4.0 Technology Programme of the German Federal Ministry for Economic Affairs and Energy*, January 2017, doi: 10.5771/9783845284651-27.

Needless to say, it is the third one that is the most controversial reality, and for several reasons. The main is that intelligent machines can be considered a wider concept which encompasses both mechanical aspects of traditional robots (and thus the ability to provoke changes in the surrounding physical environment) and a high degree of autonomy. In addition, AI is the driving force of development of machines having cognitive and decision-making capacities.
For these reasons, in this paper, I use the concept of intelligent machines, namely because of its strength and its challenging feature.

4.2. Two competing visions on humans and intelligent machines

Two competing visions (and scenarios) face each other in the current debate about future coexistence of humans and intelligent machines. Within the vast literature on the issue, it seems to me that the most significant and authoritative positions are the following two.
According to the first one *Superintelligent AI* might be the last event in human history**.** The Center for Human-Compatible AI (at Berkeley University) presents its mission in a scenario that is anything but reassuring:

> to develop the conceptual and technical wherewithal to reorient the general thrust of AI research towards provably beneficial systems […] The long-term outcome of AI research seems likely to include machines that are more capable than humans across a wide range of objectives and environments. This raises a problem of control: given that the solutions developed by such systems are intrinsically unpredictable by humans, it may occur that some such solutions result in negative and perhaps irreversible outcomes for humans. CHAI's goal is to ensure that this eventuality cannot arise, by refocusing AI away from the capability to achieve arbitrary objectives and towards the ability to generate provably beneficial behavior.[16]

Main promoter is Stuart Russell, a very well-known computer scientist who has coauthored with Peter Norvig the authoritative book *Artificial Intelligence: A Modern Approach*. Other important people sharing similar views are Stephen Hawking, Max Tegmark, & Frank Wilczek,[17] who in a commentary article on the movie *Transcendence* (Johnny Depp protagonist) evoke singularity:

> Looking further ahead, there are no fundamental limits to what can be achieved: there is no physical law precluding particles from being organized in ways that perform even more advanced computations than the arrangements of particles in human brains. An explosive transition is possible, although it may play out differently than in the movie: as Irving Good realized in 1965, machines with superhuman intelligence could repeatedly improve their design even further, triggering what Vernor Vinge called a "singularity" and Johnny Depp's movie character calls "transcendence." One can imagine such technology outsmarting financial markets, out-inventing human researchers, out-manipulating human

[16] See https://humancompatible.ai/about and http://bit.ly/3r7am1L.
[17] S. HAWKING, R. STUART, M. TEGMARK, F. WILCZEK, *Transcending Complacency on Superintelligent Machines,* in *The Huffington Post*, 2014, available at http://bit.ly/3dd0fUf. Recently published: S. RUSSELL, *Human Compatible*, Viking, 2019.

> leaders, and developing weapons we cannot even understand. Whereas the short-term impact of AI depends on who controls it, the long-term impact depends on whether it can be controlled at all.[18]

The opposite view outlines a very different scenario where the same idea of "Superintelligence" is criticized as a flawed concept.

Melanie Mitchell, a professor of computer science at Portland State University, maintains "the problem with such forecasts is that they underestimate the complexity of general, human-level intelligence. Human intelligence is a strongly integrated system, one whose many attributes — including emotions, desires, and a strong sense of selfhood and autonomy — can't easily be separated. […] the notion of superintelligence without humanlike limitations may be a myth".[19]

Roger Penrose, professor of mathematics, Nobel Prize in Physics 2020 "for the discovery that black hole formation is a robust prediction of the general theory of relativity"[20] and friend of his colleague Stephen Hawking, is even sharper and, in an interview given in May 2018, in the occasion of *AI For Good 2018 Global Summit* (Geneva, Switzerland), says that the real risk are people who think that Superintelligent AI might be the last event in human history, rather than AI per se.[21]

4.3. Intelligent machines and self-control

Hereinafter I pose the issue of self-control in this discussion context, calling *Scenario 1* that of singularity and *Scenario 2* that of flawed concept.

The question is whether the development of something like self-control (or self-domestication) is conceivable in intelligent machines.

According to *Scenario 2* this is unconceivable, because of the lack (in machines) of *emotions, desires, and a strong sense of selfhood and autonomy*. At the opposite end of the spectrum, the ideas of the proponents of *Scenario 1* would seem to leave room for this possibility. However, even to follow this idea, some important questions would arise. Self-domestication is a hypothesis for explaining the reduction of aggression in the evolution of humans: should we exclude the possibility of a reduction of their aggression?

In *Scenario 2* there is room for the opposing hypothesis of humans domesticating Intelligent machines. This might be the case of regulations of AI and AI equipped machines with the aim of preventing damages to humans and environment and increasing the level of safety. This attempt is connected with the today mainstream idea to keep humans in the loop of decision making and focusing AI on human needs and values[22]. We don't know if this is just an aspiration or a realistic

[18] THE BLOG 04/19/2014 09:14 am ET Updated Jun 19, 2014; S. HAWKING, R. STUART, M. TEGMARK, F. WILCZEK, *Transcending Complacency on Superintelligent Machines*, cit.

[19] M. MITCHELL, *We Shouldn't be Scared by Superintelligent A.I.. "Superintelligence" is a flawed concept and shouldn't inform our policy decisions*, in *NYT*, Oct. 31, 2019, http://nyti.ms/3cWx4nZ.

[20] The prize was divided, one half awarded to Roger Penrose, the other half jointly to Reinhard Genzel and Andrea Ghez "for the discovery of a supermassive compact object at the centre of our galaxy": https://www.nobelprize.org/prizes/physics/2020/summary/.

[21] See https://www.youtube.com/watch?reload=9&v=dpSpwzyO0vU.

[22] e.g. *Stanford Institute for Human-Centered Artificial Intelligence*, http://stanford.io/317ttOv: led by the well-known computer scientist Fei Fei Lin.

guide for practical decisions. Either way, it is a worthy attempt to address the problems that intelligent machines present.

4.4. Augmented human intelligence

However, there is also another way of framing the issue. We might consider what is happening today in the human-machine relationship as a new twist in the evolution of "our" self-control.

A basic idea of Scenario 1 is that intelligent machines can evolve completely independently of humans. In my opinion this is not a correct assumption. Things seem to me to be in a different way. Machines, more or less intelligent, are part of a wider human-machine scenario populated by many things unified in an essential logic of bringing technologies closer to humans, towards a progressive integration. Indeed, we have entered a phase in which technologies become closer to and interpenetrate with the human body: examples start with tech *on* our bodies (e.g. wearable devices), to tech integrated *with* our bodies, to augmented humanity (senses, cognition, motion), to invisible neural signals interface up to direct neural interfaces.

To use the jargon of one of the largest economic analysis and technology information company, one would have to say that we are already in the era of augmented humanity, where technology merges with biology to extend the physical and mental capabilities of the human body.[23]

On the academic side the Stanford University (USA) is on a similar wavelength, dedicating its Spring 2021 conference to *Intelligence Augmentation*, which is described as the way to make Artificial Intelligence not replace humans but augment their capabilities. The conference is announced with these words:

> Artificial intelligence is poised to change every sector of the economy. How do we ensure that this technology will augment, not replace, humans? During HAI's spring conference, scholars and industry professionals in the fields of healthcare, education, art, and others will discuss how AI technology can best support humans as they approach critical global challenges.[24]

If this is the case, the question becomes *who* will be the subject of self-control/self-domestication: machines alone?[25] It does not seem to be probable, as there is too much interpenetration for saying that intelligent machines will develop autonomously. Humans alone? Again, an antihistorical

[23] IDC is a leading global provider of market intelligence, consulting services and events for the information technology, telecommunications and general technology markets: see https://www.idc.com/ (visited November 20, 2020). For the vision byIBM, see Rucas, *The Fourth Platform Revolution of IBM: From Hybrid Cloud to AI*, May 26, 2020, https://www.rucashk.com/the-fourth-platform-revolution-of-ibm-from-hybrid-cloud-to-ai/ (visited November 20, 2020). See also C. WONG, *Get ready for the 'fourth platform'*, November 14, 2016, https://blog.allstream.com/get-ready-for-the-fourth-platform/ (visited November 20, 2020).

[24] See at http://stanford.io/3vQA7H6.

[25] E. FOSCH-VILLARONGA, C. LUTZ, A. TAMÒ-LARRIEUX, *Gathering Expert Opinions for Social Robots' Ethical, Legal, and Societal Concerns: Findings from Four International Workshops,* in *International Journal of Social Robotics,* https://doi.org/10.1007/s12369-019-00605-z; A.P. VARGAS, E.A. DI PAOLO, I. HARVEY, P. HUSBANDS (eds), *The Horizons of Evolutionary Robotics*, Cambridge MA, 2014. The idea of a machine able to alter its own instructions is well rooted in Alan Turing ideas: see J. COPELAND, *The Modern History of Computing,* in *The Stanford Encyclopedia of Philosophy*, 2017.

uncontaminated (by technology) humanity simply has never existed. Humans in their present situation and experience, rather resemble to persons "whose physiological functioning is aided by or dependent upon a mechanical or electronic device", which is the Oxford dictionary definition for cyborg.

In this realistic perspective, machines will not destroy humanity. Humans will co-evolve with the machines they create, which they will, one way or another, control through social, ethical, and legal rules. In addition, humans, integrated with mechanical or electronic devices, will continue their evolution by developing their self-control as cyborgs.[26]

5. Coevolution of humans-machines and our ecological niche

Machines are part of the game of action-retroaction which happens in the ecological niche, with other entities populating it such as humans (with their development of institutions as law, economic activity and exchange) and non-human animals and more.

The vision of the ecological niche allows us to have a more complex and richer vision of our future: we are not solitary agents, even if we bear higher responsibilities that are related to our development of a rich system of communication and self-control based on language and neuro-awareness (a system which does not exclude aggression might amplify in some social conditions).

How this will all work and along what specific dynamics is an agenda for a further research project. For example, under the self-control/self-domestication hypothesis, the enhancement of humans+machines should not (necessarily) serve their aggression, which might continue to decline. However, one must consider that an increase in power usually generates control problems: what could drive the evolution towards self-control or aggression? Also, to have an effective niche-building process, concrete feedback from selective pressures is needed, such as chips in the brain, more fitness for cyborgs, and more.[27]

[26] On co-evolution, see P.J. RICHERSONA, R. BOYD, AND J. HENRICH, *Gene-culture coevolution in the age of genomics,* in *PNAS*, 107, 2, May 11, 2010, 8985-8992, https://doi.org/10.1073/pnas.0914631107; O.R. GOODENOUGH, *Mind viruses: culture, evolution and the puzzle of altruism, Social Science Information*, 34, 2, 1995, 287.

[27] I am in debt with Telmo Pievani and Oliver Goodenough for their extremely kind and helpful suggestions and remarks. Of course, the final responsibility for the article is entirely mine.