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-*

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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.

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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.

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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.

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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.

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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.

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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).

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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.

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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.

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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.

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