Genetic Information: The individual, the family and the Humankind

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1. Introduction

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

2. Genetic information and the individual

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

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

3. Genetic information and the family

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

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

4. Genetic information and the Humankind

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

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

5. Conclusion

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

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

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

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

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