Genetic Data and Discrimination

CARLA FARALLI

As is known, a Human Genome Project was started in the late 1980s, which drew the public's attention to a revolution begun a few decades earlier, in 1953, with the discovery of the DNA's double-helix structure. As the name suggests, the purpose of the project was to map the human genome by describing the structure, position, and function of the genes that characterize the human species.

This research has been amplified and distorted under the pressure of great economic interests, among other factors, and has been touted as the culminating phase in the quest for the "biological Grail"[1], revealing what it means to be human, and having the potential to change our philosophical self-understanding, by showing us how life works. This research has engendered great expectations as to the use it can be put to in diagnosing, curing, and preventing many diseases, this on the basis of the assumption that every aspect of our individual and social life can be traced to our genes.

More recent studies have underscored the significant role of non genetic factors in an organism's formation and behaviour, this owing, for example, to the complexity of the processes by which genes, proteins, and the environment interact. Further, as much as genetic testing may make it possible for us to detect, before or after birth, genetic anomalies responsible for a disease now in progress or potentially in the making, and may also make it possible to locate defective genes in DNA, there is nothing like a cure in most of these cases, because there is still too wide a gap between progress in diagnosis and available therapy.

Even so, we have witnessed in recent years a great increase in the use of genetic testing, from which genetic data is extracted, and unlike any other type of personal data, this data is structurally shared, permanent, and transmissible, that is, it pertains not to a

---

[1] These words can be found in W. Gilbert, "A Vision of the Grail," in *The Code of Codes: Scientific and Social Issues in the Human Genome Project*, Cambridge (Ma), Harvard University Press, 1992. Some of the most critical commentary can be found in R. Hubbard, E. Wald, *Exploding the Gene Myth*, Boston, Beacon Press, 1993; D. Nelkin, M.S. Lindee, *The DNA Mystique: The Gene as a Cultural Icon*, Ann Arbor, The University of Michigan Press, 1995; R. Lewontin, *It Aint's Necessarily So: The Dream of the Human Genome and Other Illusions*, New York, New York Review of Books, 2000.

C. Faralli, "Genetic Data and Discrimination",
Jura Gentium, ISSN 1826-8269, XVII, 2020, 1, Homo medicus *e* commodification*. Una prospettiva bioetica*, pp. 179-186

single person but to the entire class this person belongs to, and it further makes available information not only about what we *are* but also about what we could *become*.

This data identifies for each of us our genetic makeup, which cannot be modified at any point in our lifetime: it captures something about us as unique individuals, and it uniquely relates us to other individuals; so, too, it functions as a direct biological linkup between generations, and as such it is "immortal" – it is so unlike all other biological traits, which are phenotypic (relating to an organism's physical appearance as distinguished from its genetic makeup) and accordingly become extinct when the individual dies. As Stefano Rodotà argues[2], this explains the special status and centrality genetic data has come to have within the realm of personal data.

The legal definition of genetic data in Europe can be traced to a 1997 recommendation of the European Council, No. R(97)5, stating that the term "refers to all data, of whatever type, concerning the hereditary characteristics of an individual or concerning the pattern of inheritance of such characteristics within a related group of individuals". The main point emerging from this definition is that, as was noted a moment ago, unlike all other kinds of personal data, genetic data is structurally shared, permanent, and transmissible, which entails some important consequences in regard to at least two issues, the first having to do with the fact that genetic data makes it possible to identify one's membership in a group, and the second concerning the rights of the different persons in such a group.

Our membership group consists of our biological family, which does not coincide with our legal family (thus, for example, the group does not include a spouse or an adoptive parent, though it will include gamete donors in cases involving artificial insemination, or it will include a surrogate mother giving birth on condition of anonymity). Some theorists have also gone to the extent of hypothesizing in this biological membership group the inclusion of future generations, a class that gives cause for concern from a legal and theoretical point of view, given the difficulty of regarding

[2] Stefano Rodotà was the first civil lawyer in Italy to address the question of genetic data, well ahead of his time as he anticipated contemporary legal doctrine by arguing for the exceptional status of such data. This is a view he first set out in *Tecnologie e diritti*, Bologna, Il Mulino, 1998, and then amplified in *La vita e le regole: Tra diritto e non diritto*, Milano, Feltrinelli, 2008.

as rights-holders beings who do not even have a potential existence, in that we do not know whether, and if so how, they will exist.

The issue we confront as we consider this biological membership group is that of the rights their members should be entitled to – such as the right to access information about other members in the group, or the right to privacy (which in this case includes the right not to know) – along with the related issue of how such rights should be exercised, as well as that of the authority to use and share the group's data.

As can clearly be appreciated, we have to do here not only with traditional problems –such as the protection of individual rights, the balance between individual and social interests, and the relation between the private and public sphere – but also with new problems stemming from the scientific and technological developments that have revolutionized our communications and have made it possible manipulate life in ways unthinkable only a few decades ago (examples being artificial insemination, organ transplant, and genetic engineering).

It is international law that offers the earliest examples of frameworks by which to regulate this whole area of activity in protecting fundamental rights[3]. The first such document is considered to be the Nuremberg Code, the outcome of the guilty verdict issued by the International Military Tribunal in 1947 over the course of the Subsequent Nuremberg Trials, after which came other fundamental declarations, up to the Universal Declaration on the Human Genome and Human Rights, adopted by UNESCO in 1997; the Convention on Human Rights and Biomedicine, also of 1997; and the European Union Charter of Fundamental Rights, of 2000.

The Universal Declaration on the Human Genome and Human Rights starts out in its preliminary matter

> recognizing that research on the human genome and the resulting applications open up vast prospects for progress in improving the health of individuals and of humankind as a whole, but emphasizing that such research should fully respect human dignity, freedom and human rights, as well as the prohibition of all forms of discrimination based on genetic characteristics.

---

[3] For a condensed yet accurate discussion of international law in this regard, see I.R. Pavone, "Diritti dell'uomo e genetica", in *Enciclopedia giuridica Treccani*, update vol. 15, Roma, Istituto dell'Enciclopedia Italiana, 2007. Cfr. E. Stefanini, *Dati genetici e diritti fondamentali: Profili di diritto comparato ed europeo*, Padova, CEDAM, 2008.

Having proclaimed in Art. 1 that the "human genome [...] in a symbolic sense [...] is the heritage of humanity", the document sets down in Art. 2 the principle that "everyone has a right to respect for their dignity and for their rights regardless of their genetic characteristics" and "that dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity." And Art. 6 states "No one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity". In 2003, UNESCO relied on this document for its own International Declaration on Human Genetic Data, setting forth principles for the collection, processing, use, and storage of such data, which it recognizes in Art. 4 as having a special status, for it "may have a significant impact on the family, including offspring, extending over generations, and in some instances on the whole group to which the person concerned belongs".

The Convention on Human Rights and Biomedicine, for its part, and as is known, was the outcome of a long and laborious process begun in 1991, and not until 1997 did it come into force, when numerous states signed it in Oviedo. It codifies pre-existing yet fragmentary pronouncements the Council of Europe had issued to the member states beginning in 1970 with the aim of achieving international cooperation on medical ethics, and it also codifies some (likewise fragmentary) recommendations on genetics dating to the 1980s, beginning with Recommendation No. 934 on genetic engineering.

Chapter 4 of this convention is devoted to the human genome, with four articles prohibiting discrimination in any form against anyone on account of their genetic heritage (Art. 11); allowing genetic testing only for medical purposes and for research, and only with appropriate genetic counselling (Art. 12); permitting intervention on the human genome only for preventive, diagnostic, or therapeutic purposes, and only if its aim is not to modify the genome of any descendants (Art. 13); and prohibiting the use of medically assisted procreation for the purpose of choosing a child's sex, unless such use is necessary to avoid a serious hereditary disease related to sex (Art. 14). So we have here a series of provisions affirming a right to an individual genetic identity – his by way of a corollary, as it were, of the rights to life and health. In 2008, a protocol to the convention was issued on genetic testing (its full title being "Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes"), setting forth

some fundamental principles, among which an obligation to ensure adequate genetic counselling when doing genetic testing, and the right not to be informed. Particularly interesting in this regard is Art. 13 of the protocol, introducing an exception to Art. 6 of the Oviedo convention, on the protection of those who cannot consent: the exception states that genetic testing may be carried out on someone lacking the capacity to consent if such testing is undertaken for the benefit of family members, this so long as certain conditions are met, including the condition that the benefit gained is important for the health of this person's family members, or otherwise that the test allows them to make an informed choice with respect to procreation, and that certain criteria are met ensuring minimal risk for the person subject to the test.

The European Union Charter of Fundamental Rights – signed in Nice in 2000, and now referenced in the Treaty of Lisbon, which came into force on 1 December 2009 – includes genetic features in Art. 21 in a full list of prohibited grounds of discrimination:

> Any discrimination based on any ground such as sex, race, colour, ethnic or social origin, genetic features, language, religion or belief, political or any other opinion, membership of a national minority, property, birth, disability, age or sexual orientation shall be prohibited.

The European General Data Protection Regulation (EU) 2016/679 (GDPR), unlike the "mother directive" (Data Protection Directive 95/46/EC), devotes specific attention to genetic data[4]. In the GDPR, genetic data are considered a specification of health data and are qualified on the basis of two elements: their source (they come from the biological samples extracted from a person) and their object (they contain that person's inherited or acquired genetic characteristics)[5]. In Italy, before the GDPR came into force (May 25, 2018), the legal provisions dealing with genetic data were scarce, essentially limited to Article 90 of the so-called Privacy Code (Legislative Decree No. 196 of 30 June 2003), under which the processing of such data was conditional on a specific authorization by the Italian Data Protection Authority, as well as on general authorizations by the same authority, spanning from the first such authorization of 2007,

---

[4] See Whereases 14, 35, 53, and 75 and Articles 4.1, 4.13, 9.1 and 9.4.

[5] At Article 4(13) GDPR genetic data are defined as "personal data relating to the inherited or acquired genetic characteristics of a natural person which give unique information about the physiology or the health of that natural person and which result, in particular, from an analysis of a biological sample from the natural person in question".

extended by means of subsequent yearly provisions, to the authorization of 2016, which remained in force until Provision No. 146 of 5 June 2019 ("New Prescriptions of the Processing of Particular Classes of Data"), which the Italian Data Protection Authority issued pursuant to Article 21 of Legislative Decree No. 101 of 10 August 2018, containing provisions for aligning Italian national law with the GDPR.

The special emphasis that discrimination receives in the documents just briefly discussed shows that "genetic" discrimination has now made its way into the open menu of forms of discrimination, and yet it differs from these other forms in at least two respects.

In the first place, considering the nature of genetic data as previously discussed, discrimination based on genetic features is discrimination affecting not the single individual but the biological family the individual belongs to. This biological membership group makes it necessary to carefully consider the question as to *who* makes up this group and what "rights" they each have, as concerns, for example, the ability to access information about other members of the group, or the privacy of those concerned (including under this heading the right not to be informed), and the authority to use data pertaining to the group.

Clearly, there are profound implications for our individual personality once we find out what our genetic destiny will be as revealed through a predictive test (and it should be mentioned in passing from the outset that these tests are reliable only for monofactorial genetic diseases, and that in the vast majority of cases the disease will instead be polifactorial, making it impossible to predict its onset). But in any event, the information so gained can lead us to take a preventive strategy designed to reduce or minimize the risk involved, but it can just as easily act as a source of anxiety and may even lead to depression or to tragic choices (the most frequently cited case in this regard is that of Huntington's disease, which tends to have a late onset). Whence the need to respect everyone's right to decide whether to be informed about genetic test results and what they mean. On a philosophical level, the right not to know comes into conflict with the principle of responsibility, to be sure, but it is certainly a component of the right of self-determination, or the right to freely make choices in life.

Stefano Rodotà comments in this regard that the ability to predict our biological future paves the way for greater control on our part, enabling us to freely make choices

in situations hitherto treated as necessitated, such that what is now left to chance will at some point be a matter of freedom. We are in this way working toward situations facilitating us in deliberately designing our biological future: this opens up the prospect of an "antidestiny," where the human being "governs" situations rather than sustaining them.

In the second place, genetic discrimination differs from all other forms of discrimination in that it may target an individual not on the basis of a current condition, but on the basis of the "risk" (or what is "presumed" to be the risk) that such a condition may at some point develop, even though it may not develop at all.

This is because, unlike any other kind of personal data, genetic data provides information not only about what someone "is", but also about what he or she could "become". As Rodotà comments, this expands the range of possibilities for classification by introducing concepts such as "prediction," "proneness," and "at-risk person." But these interpretive classes – consider that a cautious use of them is already being recommended even in predictive medicine – can give place to perilous misunderstandings if "tapped" from mainstream genetics and made to spill over into the realm of social policy. Indeed, we incur the risk of taking a potential condition, or worse, a hypothetical one, its likelihood often determined on the basis of statistical methods, and turn it into an unchangeable predestination, thus fostering an environment receptive to an entire spectrum of consequences, involving matters ranging from the legal treatment of those concerned to our social perception of them, and, perhaps even more alarmingly, involving the world of work and insurance, by threatening to usher in a caste society with a class of people regarded as so much a "risk" or a "liability" that they become "unemployable" and "uninsurable".

It is against the background of these potential risks that a trend has recently developed in genetic research with the emergence of so-called genetic reductionism, a sort of modern avatar of the biological reductionism championed by Francis Galton (1822-1911) and Cesare Lombroso (1835-1909). This brand of genetic research has received strong criticism, though, with a firm stand taken against it by many geneticists, not least of whom Craig Venter and Francis Collins, the promoters of the Human Genome Project, who have observed "We're clearly much, much more than the sum total of our genes." For which reason, as has been underscored by another great American geneticist,

Victor McKusick, it would be a grave mistake to think we have understood everything about ourselves just because we have sequenced our own genome, and likewise grossly misguided would be the idea that the human condition is simply a direct and inevitable consequence of our genome.

Carla Faralli
Università di Bologna
carla.faralli@unibo.it