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Personalized medicine and genetic data legislation in Italy and in the German-speaking countries

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***Abstract*— The term "personalized medicine" has been generated to indicate strategies for targetting medical treatments in each individual patient based on his/her own phenotype, genotype, lifestyle, and clinical records.**
Initially, these strategies were largely based on classical pharmacogenetic and pharmacogenomic methodologies. Later on this concept became progressively wider and included further predictive genomics approaches enabling patient stratifications based upon genetic risk to develop specific diseases.
Indeed, improved understanding of human genome brought personalized medicine into the novel dimension of "predictive medicine" which enables quantitation of disease risk in addition to disease presence.
The legal, bioethical and socio-economic implications of this advancement have only recently started to attract interest on how to compromise the need of scientific progress and the protection of the fundamental rights both at the individual and at the community levels. This is clearly a very sensitive as well as an extremely complex area as it handles genomic information potentially related to the individual fate.
In fact, in several countries, including Italy, there still is no specific discipline which addresses the important issues of genetic/genomic data protection and use. At variance, other Countries are more advanced. Germany, for instance, has a complex regulatory body, termed *Gendiagnostikgesetz* (GenDG), in force since 2010, providing detailed recommendations to protect individuals from discrimination based on their genetic characteristics, and also establishing special rules in the insurance field. Similar German law, Swiss and Austrian laws were prompted by the notion of "Genetic Exceptionalism". Indeed, both Switzerland and Austria have legislation on genetic data with interesting profiles on insurance practice.

***Keywords*— genetic data, personalized medicine, insurance contracts, legislation**

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I. Personalized Medicine and genetic testing

Current personalized medicine originates from the advancement in the field of genetics over the past century.
However, more recent progress determined an unanticipated broadening of personalized medicine perspectives. Indeed, it is becoming progressively clearer that individual "genetic disorders" may originate from mutations in single or multiple genes as well as from functional abnormalities in genomic organization such as those determined by epigenetic modifications.
Current technology now offers very powerful tools to identify this variability at the whole genome level.
From a diagnostic perspective, this novelty provides unprecedented opportunities to develop tests to identify both the presence of a specific disease and the risk of developing a specific abnormality, enabling one to focus on prevention and early intervention rather than on treatment. In many areas, the clinical interventions can be life-saving.
From a therapeutic perspective, this same technology now enables to tailor pharmacological options on the needs of the individual patients, thus replacing empirical with scientific approaches, improving efficacy and reducing side-and unwanted effects.
Completing the Human Genome Project (deconvolution of the entire human genome) has represented a milestone in the field of human genetics which fostered further and highly ambitious programmes.
A large number of genetic tests have been commercialized which enable the identification of an array of pathological traits. Some of these tests are predictive and well-suited one to anticipate the risk of developing a specific disorder before its debut. Other such tests make possible the production of a drug response profile and may help in identifying the most appropriate pharmacological treatment.
The U.K. Human Genetic Commission defines "genetic test" "a test to detect the presence or the absence of, or a change in, a particular gene or chromosome or a gene product or other specific metabolite that is primarily indicative of a specific genetic change".
Eleven categories of genetic tests are identified, three of which are, in particularly, considered "genetic test in the context of inherited or heritable disorders" which "may have important implications for the health of the person concerned or members of their family, or have important implications concerning reproductive choices".
These tests include Diagnostic, Predictive and Presymptomatic tests.

Diagnostic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms.
Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer.
Presymptomatic testing can determine whether a person will develop a genetic disorder before any sign or symptom appears. The results of those testing can provide information about a person's risk of developing a specific disorder and help in making decisions about medical care.
It must be underlined that subjects undergoing genetic tests are not necessarily „patients" as they might also be individuals requiring consulting such as relatives of affected individuals (at risk individuals).

II. Italian legislation on genetic data

The definition of genetic, pharmacogenetic and pharmacogenomic tests in the provision of extension of the Italian General Authorization to Treatment of Genetic Data by the Data Protection Authority has accepted this classification.
First of all, the Authority defines "genetic test" a clinical test on a gene or gene product or function or on other DNA or chromosome region aiming at enabling a diagnosis or confirming a clinical hypothesis in an affected individual (diagnostic test).
According to this definition, genetic test is also one aimed at proving or excluding the presence of a mutation causing a genetic disorder in an unaffected individual (presymptomatic test) or one aiming at evaluating the individual susceptibility to develop specific multifactorial diseases (susceptibility prediction test).
In addition, the provision identifies the pharmacogenetic and pharmacogenomic tests as individual measures. The former is defined a genetic test aiming at identifying specific variation in DNA sequence enabling prediction of individual response to drugs, both in term of efficacy and in term of risk of adverse effects. Pharmacogenomic tests are defined genetic tests aiming at similar objectives at the whole genome level.
Use of biological samples and of results from these tests contribute to define a "private" genome profile and must be considered very sensitive data. Accordingly, they must be subjected to an high level of protection by means of all measures necessary to prevent the violation of fundamental rights and of dignity of the owners.
Current Italian legislation mainly strengthen individual standing, generically positioning data protection within the fundamental rights of the individual, as also derived from the Italian Personal Data Protection Code (*Codice della Privacy*), which further underlines the need of human dignity to be respected in treating personal data (Sect. 2, d.lgs. 30 *giugno* 2003, n. 196).
Consistently, the Charter of Fundamental Rights of the EU (article 8) establishes the right of each individual to have his/her personal data protected. Such statement certainly applies to genetic data.
The Italian Personal Data Protection Code identifies a particularly stringent set of recommendation for sensitive data. These include medical data which also encompass genetic data. In particular, the Code establishes that processing of genetic data, shall be allowed exclusively in the cases provided for in the ad-hoc authorisations granted by the *Garante*, after having consulted with the Minister for Health who shall seek, to that end, the opinion of the Higher Health Care Council (Sect. 90, d.lgs. 30 *giugno* 2003, n. 196).
The definition of genetic data is therefore highly relevant as it establishes the appropriate levels of legislative protection to be applied.
In principle, one can envision two different conceptions of genetic data. Genetic data can be defined as all of the information relative to the genetic features of a certain individual including those related to his/her family history. A more restrictive definition strictly links genetic data to the genetic test result.
The Italian *Istituto Superiore di Sanità* accepts the wider interpretation and defines the genetic test as one which is based on DNA, RNA, chromosome, protein, metabolite analysis aiming at identifying genotypes, phenotypes, mutations related to hereditary disorders, including prenatal, neonatal or carrier screenings as well as screenings in at risk families. Tests for research purposes are excluded from the definition.
Of course, which definition of genetic data one may decide to use also affects the level of protection to which data circulation has to be subjected and impacts on the specific policies to be followed.

III. Genetic protection in European and international Law

Another definition of genetic data is contained in a Recommendation of the EU Council dated 1997. According to this definition, the term genetic data includes all data related to individual heritability, whether they relate to the transmission of a normal or a pathological trait.
In essence, they are defined as personal data specifying permanent information which are both unique of a particular subject through his/her lifespan and shared within generations, which accounts for legal significance.
Indeed, genetic data, at variance with other personal data, can be transgenerationally shared. This latter feature generates a biological link with a reference group termed biological family which may or may not collimate with the juridical family.
Whether members of the biological family also share rights regarding the access to information on other members or the right of not knowing are open issues.
The "right to know" and "right not to know" are commonly subsumed under the concept of informational self-determination. A further problem arises when the right to know of a certain individual collides with the right of a

biological relative of not to know. How the biological family member data should circulate within the biological family and what level of protection is appropriate is absolutely controvert.
Recent scientific and technological advancement has created a novel dimension in the field of human fundamental rights, widening the perspective of bioethical issues related to recognition of diversity and protection from several potential forms of genetic discrimination and positioning these „fourth generation rights" along with more traditional rights such as the right of self-determination and of not knowing. An important feature characterizing this fourth generation rights is their potential extension to future generations, a very controvert field at the moment.
According to UNESCO Universal Declaration on the Human genome and Human Rights adopted in November 1997 - which includes in its section II important substantive principles relating to bioethics, such as "Respect for human dignity and human rights (Article 3.1); Informed consent (Article 6); Non-discrimination and Non- stigmatisation (Article 11); Protection of future generations (Article 16)" - and the International Declaration on Human Genetic Data adopted in october 2003, every genetic test, i.e, including all predictive genetic tests, requires free consent based on adequate information. In addition, it requires that the patients should have a fundamental right to decide for themselves whether or not they wish to be informed of any findings.
As stated by the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine, Predictive genetic tests may be subjected to appropriate genetic counselling (article 5) and predictive tests may be performed only for health purposes or for scientific research linked to health purposes, and subjected to appropriate genetic counselling (article 12). This principle is confirmed in article 8 para. 2 of the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research, approved and ready for signature since november 2008. Until now, the Protocol to the Convention, which opened for signatures on November 27, 2008 has not yet obtained the five ratifications, including those of four Council of Europe states, necessary for entry into force.
These predictions contribute to strengthen the protection of genetic information in the context of personal information, making more cogent the statement under section 23, para. 2, Italian Personal Data Protection Code, (d.lgs. 30 *giugno* 2003, n. 196), concerning the treatment of personal data). Consent shall be given in writing if the processing concerns sensitive data (Sect. 23, para 4).
According with the main principle of the protection of human dignity and of fundamental rights, as appearing in the Charter of Fundamental Rights of the European Union, and in the Convention of human Rights and Biomedicine (article 11), between the "25 Recommendations on ethical, legal and social implications of genetic testing" of EU Commission, the recommendation n. 11, against discrimination and stigmatisation, states that "timely access to genetic testing should be based on need and appropriately resourced with no discrimination based on gender, ethnic origin, social or economic status. Personal medical data, including genetic data, must not be used in ways that disadvantage or discriminate unfairly against individuals, families or groups in either clinical or non-clinical contexts, including employment, insurance, access to social integration, and opportunities for general well-being".
The risk of discrimination and stigmatisation due the individual genetic profile is particularly significant in the area of private insurances related to risk of life or disease. Genetic tests are indeed of interest for insurance companies when adopted as tools for stratifying risk to protect from asymmetric information and to adequately adjust premium levels.
The essential problem is therefore how to compromise insurance company interests and the rights of the insured of not being discriminated. Germany has already demonstrated significant interest and activities in this area and produced a legislation termed *Gendiagnostikgesetz* which may represent a reference in this area.

IV. *Gendiagnostikgesetz*

The main aim of German legislation, (which was issued in 2010 after a long debate), is to protect individuals from discrimination based on their genetic disposition; thus, from being reduced to a genetic substrate. In the eyes of the legislators, human dignity implies the right not to know about one's own genetic dispositions. Therefore, the right to informational self-determination regarding genetic data merits protection. The general clause laid down in Sect. 4 para. 1 GenDG states that nobody shall be discriminated against because of their (or genetically related persons') genetic characteristics, the carrying-out, or refusing to do so, a genetic test or analysis on themselves (or on a genetically related person) or because of the result of such testing or analysis. This prohibition has binding effect on private individuals as well as on public entities.
For certain areas of law the statute substantiates the general prohibition by means of individual rules. Amongst those there are a number of rules on insurance, which are located in Sect. 18 GenDG.
Indeed as provided by GenDG, Genetic data is considered sensitive data because it discloses medical information about a human being with life-time significance. Thus, arguments against its disclosure are tied to human dignity. The right to informational self-determination has to be protected as a substantial part of human dignity. The right not to know about one's genetic characteristics and thus not to be forced to carry-out genetic testing has to be protected in the same manner. Moreover, in the case of insurances a full disclosure harms the system having as a consequence a too strong risk selection. Persons with genetic low risks would migrate and so the premium for the individuals with bad genetic risk profiles would rise unacceptably.
On the other hand the disclosure of genetically based risk would enhance the objectivity of the application process enabling the insurer to make a complete risk-assessment. It

would make it possible to lower premiums, because the insurer would not have to take into account an uncertainty concerning the health risk of the person to be insured and thus no safety margin for this risk would be necessary.
Both sides' argumentative approaches had been considered in the German legislative procedure and lead to a compromise in sect. 18 of the German Genetic Diagnostics Act (GenDG). It prohibits the demand to carry out genetic testing as well as the demand for results of already undergone genetic testing. But an exception of this prohibition exists for insurance contracts exceeding the value of 300.000 € or an annuity of 30.000 €.

V. Genetic data protection in Austrian and Swiss legislation

Similar German law, Swiss and Austrian laws were prompted by the notion of "Genetic Exceptionalism".
Both, Switzerland and Austria have legislation on genetic data. The former introduced it in 2004 in the form of the Federal Act on Human Genetic Testing - HGTA. However, before its introduction, Switzerland issued a constitutional law, Article 119 of the Federal Constitution, aiming to permit – only with the consent of the individual – the analysis and recording of genetic data. Austria, on the other hand, already issued the GTG (*Gentechnikgesetz*) in 1994 – which has been amended several times in recent years – that precisely delineated interesting profiles on genetic data protection.
German-speaking countries issued a special legislation on genetic data according to the principle of "Genetic Exceptionalism". Indeed the results obtained through genetic analysis are characterized by their relevance for long periods. They can be defined as personal and relevant health data that typify the identity of a person and that can at the same time reveal information about third parties (such as relatives). Indeed, a common element among German-speaking countries' regulations is the protection of human dignity.
The purpose of these laws is to prevent that the study of human genetic characteristics can be transformed into genetic discrimination, while maintaining each individual's possibility of getting tested. In order to safeguard this right, German-speaking countries have highlighted the principle of "*Beratungsvorbehalt*" or medical consulting.
These regulations prescribe that individuals consult a doctor, a geneticist, before and after performance of genetic tests. The Austrian law is much more rigid because it requires a special form of consultation. After medical consulting, a report is to be written and afterwards signed by the user. To avoid pressure during decision-making in relation to the use of genetic results, the user is never mentioned as "patient" but rather as "interested person" or "consultation's user". Moreover, the term "danger" is never used but rather the term "possibility" in order to let the person decide freely whether to perform the analysis or not. The Swiss regulation, as well as the German one, pays more attention to the "*Bedenkenzeit*", which is the time that a person must have to think over his decision before performing genetic analysis. To ensure the quality of the consultancy, "*genetische Betreeung*" has been developed. It gives users a proper assistance in the most impartial way. Furthermore, the legislation of these countries has provided the "*Arztvorbehalt*", the principle of medical reserve. Not all doctors can perform genetic analysis or carry out consultations, only geneticists.
Another important aspect, on which these regulations focus, is the consent or "*Einwilligung*". Consent plays a significant role, because it embodies the will of the person to find out about his or her genetic data and for this reason, it must be given in written form. Nevertheless, any person who changes his mind also has a possibility of revocation, because of "*Recht auf Nichtwissen*", the right not to know. In German-speaking countries there is an "*informationelle Selbstbestimmung*", a person's self-determination in deciding whether or not to obtain of certain data. An important difference in the regulation of these countries regards data disclosure. While Austrian legislation is similar to the German one, Swiss law provides in sect. 19 the possibility for the doctor to ask a competent authority to be released from his duty to medical confidentiality and be able to disclose data to protect a "higher" interest, such as that of partners or family members. In order to reach a decision, the authority may be assisted by a geneticists' committee.
A peculiarity of Swiss legislation is the disclosure to third persons. Moreover, the doctor can decide to show the results to other persons if he obtains permission from a committee, even if the individual refuses disclosure.

VI. The impact on insurance practice

The use of genetic information in insurance has been identified as a key problem. The line between the right of insured persons not to disclose their genetic data and the insurers' interest in obtaining such information has led to difference in the legislations of both German-speaking countries. While the disclosure of genetic data is possible under certain conditions in Switzerland, it is completely banned in Austria.

A. *Swiss legislation*

Swiss law dedicates the fifth chapter of the Federal Act on Human Genetic Testing to insurance practice. It stipulates that insurers are forbidden from demanding genetic tests as a condition for granting an insurance contract, but are allowed to ask for existing genetic information, if the sought insurance coverage exceeds a certain amount.
Insurance providers may not require either pre-symptomatic or prenatal genetic tests prior to providing insurance. For life insurance with an insured sum up to a maximum of CHF 400,000, or voluntary invalidity insurance with an annuity not exceeding CHF 40,000, insurers cannot require the disclosure of genetic results or genetic tests. The possibility of splitting contracts is banned. Therefore, if an individual signs several life insurance contracts, the maximum amount stipulated is

valid for the sum of the policies. The applicant must provide the insurance provider with the relevant information.

B. *Austrian legislation*

Regarding the Austrian legislation, the central provision of the law establishes that nobody shall be discriminated based neither on genetic testing or analysis of his genetic data nor that of a genetically related person or due to the result of such testing or analysis. Therefore, insurers have no access to such tests. Moreover, sect. 67 of GTG establishes that insurers including authorized representatives and co-workers are forbidden from collecting, demanding, accepting or in any way using results from their clients' genetic tests or people who want to take out an insurance policy. This prohibition also extends to the demand of delivering or accepting bodily substances for genetic test purposes.

The issuing of sect. 67 was motivated by legislator's intention to protect the information self-determination right according to the art. 8 of European Human Rights Convention. Hence, Austrian law is criticized in insurance practice. This provision banned completely the possibility to use genetic data. No exception is authorized, and even if an insurance client wants to disclose his results it is forbidden. The question regarding prior illnesses is very controversial. While in Germany sect. 18 of GenDG states that illnesses and prior illnesses may be disclosed to the extent that secs. 19 through 22 and 47 of the Insurance Agreement Act apply, in Austria this is not the case.

The Austrian provision does not refer to other regulations. Moreover, it is noteworthy that for medical underwriting, before signing an insurance policy, the client has to reveal all information that may be of relevance. This provision includes prior illnesses.

VII. Conclusion

Although discrimination is regulated at the European level, there is no regulation connected to discrimination based on genetic data. Indeed, the EU-treaty includes measures against discrimination based on gender, age, disability; but there is no mentioning of genetic diagnostics.

German-speaking countries act as "first movers" and represent a good example of permitting genetic test whilst safeguarding and protecting human dignity by avoiding discrimination based on genetic data. Indeed, people should not be treated worse because their genetic code is considered less favourable. The above mentioned legislation has shown its prerogative to promote genetic data protection, which is included as a part of human dignity in the fundamental human rights. Furthermore, these countries took into account that medicine is an ever changing discipline, therefore their laws mandate that expert commissions keep up with scientific progress through continuously updating the guidelines.

The legislations have accepted the so-called principle of "Genetic Exceptionalism" for preventing discrimination on genetic data, which could lead to Eugenics. In conclusion, German-speaking countries legislation protecting the principle of human dignity through trying to keep human beings from being reduced to a genetic substrate, represents a "best practice" regulation for such countries, as Italy that still have not issued a legislation on this topic.

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