Abstract: Responsibility means responding to the damaging consequences of technical work and in this binding perspective the general principles of guilt in genetic diagnostics and related activities are not different from any other medical performance.

Performing a genetic test however, especially when it has predictive characteristics, offers absolutely peculiar technical deontological issues. It is not and should not be considered as a mere habitual laboratory test but as a complex set of interactions that presupposes adequate information as a valid consensus to formalize absolutely in written form.

Keywords: Genetic testing; professional responsibility.

1 Genetic testing and professional responsibility

The fundamental ethical issues triggered by genetic testing are linked with medico-legal problems [1]. It has been shown that compared to traditional medicine, genetic progress is marked by an increase in uncertainty over the outcome.

This implies an amplification of the risk margin that the patient must in any case have to face and should be made aware of.

It results from a difficult task of information because of the fact that the doctor is probably not prepared with the possibility of incomplete information [2] and with significant repercussions on the patient who may not fully understand the extent of the genetic indication [3].

On these grounds, it will inevitably have to consider the benefit / damage ratio triggered by the practice of genetic testing as well as the uncontrolled diffusion of well-known “diagnostic packages” that seem to thrive precisely on the lack of knowledge that comes with adequate information.

The prescription of genetic tests and the knowledge of their findings can in fact expose the subject to stress, social alienation, and difficulties in relationships. Many people will enter the “Un-patient class”: individuals that are not cared for and therefore have no illness but are not exempt from medical testing as a portion of them could manifest psychosomatic symptoms.

Genetic compatibility is also required in organ transplants [4].

It is also necessary to consider that a genetic test provides risk indications not only for the individual but also their family members, and this raises delicate issues regarding extending the information even independently of the will of the righteous person. These problems are worsened by the limits of the test with predictability values that are not even homogeneous even within the same family group [3].

The protection from particularly sensitive figure such as that provided by genetic testing is a delicate matter but in principle any information on a person’s genetic characteristics will constitute a confidential matter that could only be disclosed by permission of the person. Anyone who receives this information should adequately protect them.
Parallel to the right of information of the person concerned is the configuration of the already-called “right to not know” or the right of an individual to not know or ignore the genetic information that concerns them even when they come from examinations carried out on family members and this is stated in an absolutely meaningful way especially in those cases where there is no therapeutic remedy for the disease in question.

In this regard, Article 10 of the Convention on Human Rights and Biomedicine of 1997, ratified into the Law of 28 March 2001, states that every person has the right to know all the information gathered about their health and that the will of a person not to be informed must be respected [5].

In the Universal Declaration on the Human Genome and Human Rights adopted by the UNESCO General Conference in 1997, the right of the subject to decide whether or not to be informed on the results of a genetic test is identified [5].

A proper genetic investigation never ignores proper genetic counselling, with complete and neutral directives.

Genetic counselling is therefore a complex but indispensable task, to be carried out both at the initial preparation stage and in the final assessment test. This counselling has its purpose in defining the risk of recurrence in the individual, in family prevention, in the possible interventions to be made. Bearing in mind that for the relationship with the patient in the genetic field it becomes necessary to address the long-term chronological reference that may exist between the diagnosis and the possible onset of pathology [6].

With these assumptions, it is entirely possible to assume that professional responsibility in the genetic field is in fact the basis of assessments also of a legal medical nature dissimilar from those ordinarily in use for the usual surgical medical activity.

The activity required for “genetic counselling” is to be understood as an extended commitment to all that comes after the test execution, ranging from the indication of the survey to full information and accurate verification and interpretation of results.

In such a context, the possibilities of error are practically infinite, as are the consequent assumptions of professional responsibility.

One of the most debated issues is that of a missing or incorrect prenatal genetic diagnosis [7] or misinformation of the mother followed by an unwanted birth with the impossible imposition of compensating the “birth damage” as well as the interest not to be born [8].

In Italy, the issue was resolved by a ruling of the Supreme Court (Third Judgment No 16123 of July 2006) whose fundamental passage states that there is no configurable right not to be born. Another important aspect for the Supreme Court is that related to Law 194/78 [9]. It states that the legal possibility of interrupting pregnancy in the second trimester is motivated not by the presence of serious fetal malformations but by the fact that such malformations have or not determined an actual danger to the mother’s psychic health, with the definitive exclusion of so-called eugenic abortion [3].

The ongoing problem is the nature and limitations of the obligations of carrying out a genetic investigation with the debate on the “obligation of means” or “obligation of outcome” of medical services with a general favour for the first hypothesis. This is motivated by the finding that the medical service has increasingly assumed a competitive nature having the same purpose as being the result of a complex pool of activities none of which can be regarded as an autonomous service.

For a genetic test with such a request for technical skill and accuracy at least for the most frequently investigated illnesses, the legal configuration of an obligation is the result while it will be a requirement of means in the case of rare genetic diseases.

In the case of rare genetic diseases, underlined by the guidelines of the ISS, the low expected frequency of delivery often renders inadequate the validation of the genetic test and insufficient if it does not contradict the information. This necessitates that “medium diligence” should require less selective criteria than those imposed by more routine tests. [10] Such a distinction is too constrained and does not find real justifications to exist precisely in the motives that should support it. In any case, it will be necessary to consider, on the one hand, the preliminary determination of the subject matter of the contract (adequate information) and, on the other hand, the scientific proposition of a test developed in research activity and validated for its execution in diagnostic services.

The interpretation of the medium diligence standard must be considered with greater attention, as it corresponds concretely to the utmost diligence required by the provisions of article 1176 of the civil code according to which diligence must be assessed with regard to the nature of the activity exercised.

The jurisprudence has for a long time also identified, in accordance with Article 2236 of the Civil Code, the identification of a special difficulty where, if the service implies the solution of technical problems of particular difficulty, the provider does not respond except in the cases of fraud or serious guilt [11].
For the application of genetic tests on rare diseases, this seems to be a non-viable hypothesis, lacking the fundamental moment of the technical problem of considerable difficulty and, where there is an uncertainty of availability and insufficient validation [3].

Moreover, performing a genetic test systematically presents the peculiar aspects of a complex and complete set of interactive moments so the test is rightly in that narrow group of medical performances in which the activity is not just one of the means used to achieve results but it is the result.

The genetic test therefore cannot be seen solely as a laboratory examination and genetic counselling but it should be understood as a partial part of a wider diagnostic curative activity with an interpretation that could even turn towards an obligation of means, to consider the fact that at each stage it is not the result but an intermediate stage [12].

Naturally, in the context of particular circumstances, those situations of actual exceptions to the reliability of the test should be adequately considered, as the correct result of the investigation must necessarily be part of a broader interpretation process with the consequent reference to the obligation of means.

Another issue regarding compliance with the guidelines of the 2004 state-region agreement has produced a uniform national arrangement of the organizational model of genetic activity. These normative assumptions referring to the structural, organizational and technological requirements of healthcare establishments constitute a new tool for assessing professional guilt hypotheses, other than classic generic blame, with a view to the contractual responsibility of the now fully established structure [13].

It is obvious that the content of the state-region agreement must be considered as binding for all medical genetics services and that their violation implies automatically also non-compliance with the law so in the case of patient’s damage any default must be estimated to be able to engage in liability profiles for specific fault due to non-compliance with regulations or orders. Moreover, as evidenced by the European Commission, there are still significant problems of technical errors from inadequate quality standards and not uniformly fixed issues that further amplify the structures of medical genetics where the boundary between technical and clinical activity is even more lenient [13].

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References

[1] Conti A., Delbon P., et al, Informed Consent when taking genetic decisions, med.legaw, 2, 337-353, 2004
[2] Di Lorenzo P, Casella C, Capasso E, Conti A, Fedeli P, Policino F, Niola M. The central importance of information in cosmetic surgery and treatments. Open Medicine. 2018, 13(1):153-157
[3] Conti A., Delbon P., et al, I test genetici, Giuffrè Editore 2007
[4] Graziano V, Buccelli C, Capasso E, De Micco F, Casella C, Di Lorenzo P, Paternoster M. Suggestions on how to make suboptimal kidney transplantation an ethically viable option. Open Medicine. 2018, 11(1):523-529
[5] Declaration on the Human Genome and Human Rights (11 November 1997) – www.unesco.org
[6] Garante per la protezione dei dati personali, 2014, General Authorisation No. 8/2014 for the Processing of Genetic Data Published in Italy’s Official Journal No. 301 of 30 December 2014
[7] Casella C, Capasso E, Terracciano L, Delbon P, Fedeli P, Salzano FA, Policino F, Niola M. Ethical and legal issues in gestational surrogacy. Open Medicine. 2018, 13(1):119-121
[8] Paternoster M, Saccone G, Marotti GM, Bianco C, Casella C, Buccelli C, Martinelli P. Ethical challenges in pregnant women with brain injury. Journal of Maternal-Fetal and Neonatal Medicine. 2018, 31(7):2340-2341
[9] Piras M, Delbon P, Bin P, Casella C, Capasso E, Niola M, Conti A. Voluntary termination of pregnancy (medical or surgical abortion): Forensic medicine issues. Open Medicine. 2016, 11(1):321-326
[10] www.iss.it/tege - Istituto superiore di Sanità
[11] Polistena A, Di Lorenzo P, Sanguinetti A, Buccelli C, Conzo G, Conti A, Niola M, Avenia N. Medico-legal implications of surgical errors and complications in neck surgery: a review based on the Italian current legislation. Open Medicine. 2016; 11: 298-300
[12] Falco G, Rocco N, Bordoni D, Marano L, Accurso, A., Buccelli, C., Di Lorenzo, P., Capasso E, Policino F, Niola M, Ferrari G. Contralateral risk reducing mastectomy in Non-BRCA-Mutated patients. Open Med. 2016; 11: 238-241
[13] Presidenza del Consiglio dei Ministri – Conferenza Permanente per i rapporti tra lo Stato, le Regioni e le Province Autonome di Trento e di Bolzano, 2004