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Antenatal genetic diagnostics in the context of genetic counselling: a medical, ethical and legal perspective in Europe

Abstract

There is no doubt that the progressive evolution and development of genetic analyses that allow the early detection of the existence of a genetic disease or anomaly, has led to a new approach to medicine: personalized, predictive and preventive. This paper aims to show European –Spanish- elements that characterize this new form of medicine in comparison with the prevailing paradigm until recently of curative medicine and will describe the ethical and juridical problems involved genetic information in the context of the genetic counselling process.

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Keywords: genetic counselling, preimplantation diagnosis, prenatal diagnosis, preconception diagnosis, medical liability

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Aitziber Emaldi Cirión,[1] Emilio José Armaza Armaza[2]

[1]Department of Biomedicine and Law, University of Deusto, Spain. Researcher: European Documentation and Research Center. University of Pau and Pays de l´Adour, France

[2]Department of Criminal Law, University of Deusto, Spain. Researcher: European Documentation and Research Center. University of Pau and Pays de l´Adour, France

Correspondence: Aitziber Emaldi Cirión, Department of Biomedicine and Law. University of Deusto, Spain, Tel +34 944 13 90 00, Email aitziber.emaldi@deusto.es

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Introduction and concept of genetic counselling

First of all, we must start from the provisions of the European Convention on Biomedicine, which stresses that tests that are predictive of genetic diseases or that make it possible to identify the subject as a carrier of a gene responsible for a disease, or to detect a genetic predisposition to a disease, may only be carried out for medical purposes or medical research and with appropriate genetic counselling (Art. 12).

We can refer to the definitions provided in Spanish Law 14/2007, of 3 July, on Biomedical Research, which states that a genetic analysis is the procedure aimed at detecting the presence, of one or more segments of genetic material, including indirect tests to detect a gene product or a specific metabolite that is indicative of a specific genetic change (Art. 3). On the other hand, genetic counselling is defined as "the procedure to inform a person about the possible consequences for his or her offspring of the results of a genetic analysis. The advantages and risks and, where appropriate, to advise him or her on the possible alternatives resulting from the analysis. It takes place both before and after a genetic test (Art. 3).

In other words, the essence of genetic counselling lies in communicating the relevant genetic facts and the reproductive options for families to plan their reproduction. This idea is regulated in Article 55 of the Biomedical Research Act, which states: when a genetic analysis is carried out for health purposes, the person concerned must be guaranteed appropriate genetic counselling. Genetic counselor, shall provide appropriate information and counselling regarding both the significance of the resulting genetic diagnosis and the possible alternatives that the subject may choose in the light of the genetic diagnosis.

One of the central elements guiding genetic counselling is that it should not be directive. Non-directive counselling promotes the autonomy or self-determination and personal control of the person seeking counselling, although this can sometimes be seen as unhelpful counselling.

Phases of genetic counselling

The process of genetic counselling in the context of reproduction has different phases that we will develop below:

First, the genetic counsellor informs people with reproductive problems about the possibility of undergoing a series of predictive genetic tests to determine the possible risks of transmitting certain anomalies or diseases to their offspring. Depending on the purpose of the tests, they can be classified as follows:

Preconceive testing: performed on the subject prior to conception to check for possible risks of conceiving a child with genetic, hereditary or chromosomal diseases or abnormalities.

Pre-implantation genetic tests

These are intended to detect possible chromosomal anomalies or genetic alterations that the in vitro pre-embryos may have before being transferred to the woman. This diagnosis is applied with the intention of having a pregnancy free of anomalies, although we must not forget that even when none are detected, there may be sporadic genetic mutations that produce the development of a fetal anomaly. Preimplantation diagnosis reduces the prevalence of certain diseases by eliminating embryos with abnormal genes.

These pre-implantation diagnostics present a problem from a legal-ethical point of view. Indeed, the problem that arises here is to determine to what extent pre-implantation diagnostics can be used to detect diseases or even mere predispositions to certain diseases Then it is necessary to decide not to transfer to the woman those pre-embryos analyzed in which these diseases have been detected.

Another option after the diagnosis, is to transfer those pre-embryo depending on the seriousness of the disease detected, its possible treatment, the time at which the disease may appear - early or adult age -, etc. Therefore, depending on the use of these diagnoses, we can speak about a new eugenics.

From the juridical point of view, Spanish Law 14/2006, on assisted human reproduction techniques (art.12) and Spanish La won Biomedical Research 14/2007, stipulates on Preimplantation Diagnosis, the following:

I. Authorized centers may practice preimplantation diagnosis techniques in the following cases:

(a) The detection of serious hereditary diseases of early onset which are not susceptible to postnatal curative treatment according to current scientific knowledge. The purpose is to carry out an embryo selection of unaffected pre-embryos.

(b) The detection of other alterations that may compromise the viability of the pre-embryo. The application of pre-implantation diagnosis techniques in these cases shall be reported to the corresponding health authority, which shall inform the National Commission on Assisted Human Reproduction.

II. The application of pre-implantation diagnosis techniques for any other purpose not included in the previous section, or when they are intended to be used in combination with the determination of the histocompatibility antigens of in vitro pre-embryos for therapeutic purposes for third parties. In those cases, it shall require the express authorization, of the corresponding health authority and a favorable report from the Spanish National Commission on Assisted Human Reproduction, which shall evaluate the clinical, therapeutic and social characteristics of that case".

Prenatal tests

All prenatal actions aiming at the diagnosis of a congenital defect, understood as any morphological, structural, functional or molecular developmental abnormality present at birth, whether inherited or not, single or multiple.

The subject of prenatal diagnosis is the fetus, although it may indirectly serve the parents, since the resulting information may determine that one of them is a carrier of a deleterious gene transmitted to the fetus. With regard to the advantages of undergoing prenatal diagnosis, are the following ones:

a. It can be used to plan the birth according to the malformations suffered by the fetus.

b. It allows medical treatment to be carried out during pregnancy.

c. It provides information on the optimum state to provoke a premature birth, or to proceed to a caesarean process.

d. It confirms the normality of the fetus, calming the anxiety of the parents, if they are a high-risk couple.

e. Provides information on possible fetal malformations leading to a termination of pregnancy for eugenic indications, within the time limits established for this purpose and whenever permitted by law.

f. Verifies fetal disability or disease, and makes it possible to assume the birth of that child with anomalies.

g. It warns about the existence of malformations that would have gone unnoticed in the neonatal period.

It is also important to state the indications for prenatal diagnosis

a. When the woman was over 35years old.

b. If the man is over 55years of age.

c. If there have been repeated miscarriages in the past.

d. If there are relatives with a history of a certain disease or disability.

In the event that these indications are not given, it would be advisable for the health professional to consider the possibility of depriving them of such a practice, if it is a prenatal diagnosis carried out with an invasive technique (amniocentesis, chorionic villus sampling, fetal blood, among others).

Postnatal tests

Tests to which newborns are subjected in order to demonstrate the possible anomaly or disease that they present; their predisposition towards a specific disorder or, finally, to confirm that they are not carriers or sick. Take, for example, the test for phenylketonuria, because with a dietary treatment rich in phenylalanine would correct the disorder.

The second phase consists of the voluntary submission of the patient to the relevant genetic tests - preconceive, pre-implantation, prenatal or postnatal. Compulsory submission to any test is not possible in Europe.

The third phase corresponds to the genetic counsellor, as it is he/she who has to study the results obtained from the tests, interpret them and assess them in order to find a correct diagnosis. Furthermore, as stated in the Biomedical Research Act, the professional who carries out the genetic counselling must offer adequate information and advice on the possible alternatives (Art. 55.2).

Finally, the fourth phase would be the choice to be made by patients from all the alternatives proposed by the genetic counsellor.

Alternatives offered by the counsellor as a result genetic diagnosis

Preconception diagnosis

After carrying out a preconception tests to find a specific diagnosis, the result may be as follows:

A. When no abnormality is detected: Pregnancy will be sought.

B. When a possible transmission of an abnormality or disease is detected, the following options are available:

i. Resorting to assisted reproduction techniques by seeking gamete or embryo donors;

ii. Resorting to either male or female sterilization to avoid having offspring.

iii. Resorting to alternative and temporary methods of contraception.

iv. Assuming offspring with abnormalities or diseases.

Preimplantation diagnosis

If pre-implantation diagnosis tests are carried out, they are:

A. When no abnormality is detected: Pregnancy will be sought.

B. When a possible transmission of an anomaly is detected, the following options are available:

i. Carrying out gene therapy on the embryo, uncertain but of future potential.

ii. Selecting pre-embryos, discarding those that have an anomaly or disease.

iii. Rejecting the implantation of pre-embryos for procreation.

iv. Carry out a prenatal diagnosis confirming the anomaly or disease and proceed to terminate the pregnancy, complying with the requirements established by law for this purpose.

v. Selecting pre-embryos for therapeutic purposes for a third party.

vi. Sex selection for therapeutic purposes. Regarding sex selection, Article 13 of the Biomedicine Convention states: "No sex selection. The use of techniques of medical assistance in procreation to select the sex of the person to be born shall not be permitted, except in cases where this is necessary to prevent a serious hereditary sex-linked disease".

The Biomedicine Convention, does not appear to prohibit sex selection when its aim is to:

A. To prevent the birth of a person suffering from a serious hereditary sex-linked disease.

B. To prevent a person from being born "carrier "of a disease even if they do not develop it.

There would be two healthy embryos - male and female - since the male would not transmit the disease to his offspring and the female, on the other hand, being a carrier, would transmit the disease to her offspring. Male sex selection could be allowed to prevent the birth of a healthy female carrier.

Prenatal diagnosis

If prenatal testing is performed, the alternatives following the results may be as follows:

A. When no abnormality is detected: the pregnancy will continue.

B. When a possible transmission of a disease or abnormality is detected:

i. Recourse to abortion in the cases permitted by Organic Law 2/2010, on Sexual and Reproductive Health and the Voluntary Termination of Pregnancy.

ii. Possible fetal therapy.

iii. Assuming an offspring with a disease or anomaly.

Postnatal diagnosis

After postnatal diagnostic tests have been carried out, the alternatives will be different:

A. When no anomaly is detected: the newborn is not affected.

B. When a possible transmission of an anomaly or disease is detected:

i. Therapeutic measures.

ii. Euphuistic or dietary treatment.

Medical liability during the process of genetic counselling

The liability of health care professionals when their negligent behavior could lead to remediable damage will be analyzed below.

Negligence in offering the different health services.

Firstly, when health professionals do not advise patients with reproductive problems of the convenience of undergoing certain genetic tests, they may encourage them to give up the procreation of children, who would probably be healthy. In this case, people would be deprived of the subjective right to procreate recognized in the Spanish constitutional

Secondly, when genetic counselor doesn´t inform about the possibility of undergoing certain predictive genetic tests, prevents the pregnant woman from knowing about certain fetal pathologies and, therefore, would limit her possibility of terminating the pregnancy under the eugenic indication when it is within the time limits established by law.

Negligence in taking samples

Once the couple has been offered to undergo certain tests - antenatal, prenatal or postnatal - to obtain a diagnosis, it may happen that due to the lack of diligence of the health professional in carrying out the intervention, a series of injuries may occur: to the fetus, to the mother or to both. In extreme cases, this could even result in death. In such cases, the health professional would be criminally liable according to the specific case.

A. Fetal Injuries

Recent advances in medicine in the field of assisted reproduction has highlighted the vulnerability of the unborn.

In view of these new forms of aggression on the embryo or fetus that threaten its integrity, including both physical or corporal integrity and physical or psychological health, the need for its protection arose and the 1995 Penal Code introduced the crime of "injury to the fetus" (art)157 and 158. It is possible that actions on the fetus are carried out not only due to professional imprudence or negligence - fetal injury during the taking of samples for predictive diagnostics - but also maliciously - injuries caused to the unborn child as a consequence of experimental activities. According to Spanish law, the human being, at all stages of development, is deserving of criminal protection, so that not only his or her life, but also his or her health and integrity must be protected. Therefore, prenatal injuries with prenatal and postnatal consequences are punishable.

B. Imprudent abortion

During the process of genetic counselling, there may be specific cases in which gross negligence on the part of the health professional causes a miscarriage - for example, when performing certain invasive prenatal tests.

Another case of abortion that may arise during genetic counselling is that based on an erroneous prenatal diagnosis. In fact, it is possible that there is an error in the opinions of the two specialists in charge, appreciating the concurrence of serious fetal anomalies that do not exist in reality (false positive diagnosis), a diagnosis that will serve to carry out a subsequent abortion. The diagnosis will be used to carry out a subsequent abortion.

C. Injury to the pregnant woman

When invasive techniques are used to take the necessary samples for the purpose of genetic analysis, injury to the woman may occur.

D. Death of the pregnant woman

Exceptionally, it may happen that the taking of samples may result in the death of the woman. In effect, this could lead to the offence of homicide.

In short, when during the course of the relevant genetic counselling tests the death of the woman is caused by professional negligence, such an act will be punishable - even if the woman consented to the medical act - when the serious disregard for objectively due care is demonstrable.

Negligence analyzing the results.

The genetic counsellor has the test results. At this point, he/she has to carry out the assessment and interpretation of the results. It is possible that the health professional may arrive at a wrong diagnosis with very different consequences depending on the stage of the process. We can refer to two different scenarios: when it is a false positive and when it is a false negative.

A. False positives

False positives are cases where the individuals tested are initially identified as having the abnormality or disease but are later found not to have it.

Based on these false-positive results, individuals make reproductive decisions that, in some cases, may have far-reaching legal and personal implications:

One scenario would be the use of eugenic abortion when it is believed that there are fetal malformations, detected, for example, by prenatal diagnosis - an erroneous belief caused by an incorrect diagnosis.

Another case would arise when, after an erroneous preconception diagnosis. It is confirmed that the future parent is a carrier of a serious genetic disease and therefore it is absolutely certain that he or she will transmit it to his or her offspring. In the event that the couple decides not to have children, the consequences can be more serious, as there are many possibilities to avoid this, including, for example, recourse to sterilization.

B. False negatives

Those falsely identified as negative are cases where the initial test fails to detect carrier status and individuals are assumed to be free of abnormalities or disease when, in fact, they are heterozygous carriers.

In this context, civil claims that would be brought against doctors - and sometimes against public authorities - for negligent behavior must be addressed. In these claims, parents seek compensation in their own name (wrongful birth) or on behalf of their child (wrongful life). If abortion were not permitted under any circumstances, these claims would be meaningless, because the error in diagnosis would not result in a deprivation of the right to terminate the pregnancy.

a) Wrongful life actions

This is an action brought by a child born with a disease or abnormality against a doctor who acted negligently. The child does not allege that the negligence of the medical staff was the cause of his injury or illness, but that the negligence led to his birth. The child is not claiming for living, but for living with a disability, so the damage is life in such a dismal situation.

b) Wrongful birth actions

This is a lawsuit brought by the parents of the defective child against the doctor. The doctor is liable for damage by failing to detect or warn the pregnant woman of the illness or anomaly suffered by her fetus in time for her to be able to have an abortion under the law.

In these cases, compensation would be claimed for the damage consisting of economic damage, derived from the expenses involved in the care, attention and medication of a child suffering from an illness or anomaly and in the moral damage produced by the affliction suffered by the parents for having a seriously ill child,

Breach of the duty to inform

The health professional's duty to inform is closely linked to the patient's right to be informed. The person shall be provided in advance with adequate information on the purpose and nature of the intervention, as well as its consequences and risks

The content of the information to be transmitted to the patient includes an explanation of the significance of the resulting genetic diagnosis and the possible alternatives available to the subject in the light of the diagnosis. Furthermore, in this field of study, it must be taken into account that genetic information is provided which, by its nature, represents a sensitive type of information, as it not only reveals information about the individual to whom the analysis is carried out, but also provides information to family members or their offspring. Moreover, the individual has the right to know or not to know whether or not he/she is a carrier of a genetic disorder.

Finally, when we refer to the liability of the professional for breach of the duty to inform, it must be proved that there is a causal relationship between said breach and the damage produced.

Disclosure of data to third parties without the patient's consent

Medical professional secrecy must also be maintained throughout the process of genetic counselling in order to safeguard the right to personal privacy, which is protected in different parts of the legal system.

Confidentiality and secrecy are based on the value of respect for individuals and their right to privacy. However, in the case of genetic testing, the results may have a potential implication for other family members. An ethical dilemma arises when the person who has been tested refuses to inform family members, who are also at risk, of the results of his or her genetic test.

The shared nature of this data with the biological family and the reproductive implications it could have, raises the question of whether third parties have rights over the data.

In this regard, it must be understood that the status of biological subject does not legitimize access to shared genetic data. Consequently, the blood relatives of a patient who is recommended to undergo a genetic analysis, who share this risk, do not have the right to be informed of this circumstance.

There is not a right to information about the genetic risk that would allow the data subject's privacy to be violated, derived from the right to protection of health. In these cases, while communicating information about a genetic anomaly to the persons with whom the genes are shared is an act of great ethical value

If the patient does not wish to inform his or her relatives of the genetic information which is relevant to their health, and the omission of genetic information would result in harm, the right to health mustn't prevail over confidentiality.[1-19]

Conclusion

As a result of the study, we have been able to verify that in the field of biological, biomedical and technological sciences, advances are evolving faster than the establishment of the ethical and legal parameters that must accompany them in order to avoid abuses

contrary to human rights and, where appropriate, to the fundamental rights of the individual. Therefore, due to this unstoppable progress, the legal system as a whole is faced with new perspectives that were hitherto unregulated and even unimagined, with the aim of resolving the different conflicts of interest that arise, ranging from that which may arise between mother and child when the former undergoes a series of predictive tests for the benefit of her child, to that which arises between the duty of healthcare professionals to keep known information confidential versus their duty to disclose information.

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Conflicts of interest

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References

1. AAVV, Hacia una nueva Medicina: consejo genético, Spain: Comares Editorial; 2013.
2. AAVV, Retos éticos y necesidades normativas en la actividad asistencia. En medicina personalizada de Precisión, Spain: Ed. Fundación Instituto Roche; 2018.
3. Emilio José AA. Criminal Protection of Public Health in the Field of Biological Emergencies: The Case of the Spanish Legal System. *Ethics and Law for Chemical, Biological, Radiological, Nuclear & Explosive Crises.* 2019;20:199–209.
4. Elena AM. Algunas consideraciones sobre la protección de datos en el tratamiento de muestras biológicas y datos de salud. *Ius et Scientia*. 2017;3(2):14–36.
5. Vicente BC .La revolución de la edición genética mediante CRISPR–Cas 9 y los desafíos éticos y regulatorios que comporta. *Cuadernos de* Bioética. 2016;XXVII(2):223–239.
6. MONTALVO JF, *Menores de edad y consentimiento informado*. Spain: Marcial Pons; 2019.
7. Leyre EU, "El daño en las acciones de wrongful birth y wrongful life", *REVISTA DERECHO Y SALUD*. 2010;19:Extraordinario–2010.
8. Aitziber EC. Legal implications of Genetic councelling / El consejo genético y sus implicaciones jurídicas. Spain: Comares Editorial; 1999.
9. Aitziber EC. "Diagnóstico prenatal", *Enciclopedia de Bioética y Bioderecho*. Spain: Comares Editorial; 2010.
10. Aitziber EC. "Diagnóstico preimplantatorio", *Enciclopedia de Bioética y Bioderecho*. Spain: Comare Editorial; 2010.
11. Aitziber EC. Surrogacy in Spain and the proposal of a legislative change for its regulation. A global phenomenon in Europe. *Revista de derecho y genoma humano: genética, biotecnología y medicina avanzada*. 2018:49(Julio–Diciembre):75–99.
12. Aitziber EM. Antenatal Genetic Diagnosis and the New Eugenics: European vision from Medical, Ethical and Legal Perspectives. *Global Journal of Medical Research*. 2022;22(6):58–68.
13. Julio César GC. Sentencia del Tribunal Supremo de 6 de julio de 2007: Responsabilidad médica. Acciones de wrongful birth y wrongful life. Omisión de información sobre la conveniencia de someterse a la prueba del triple screening a gestante que no presentaba factores de riesgo relevantes. Nacimiento de niño con síndrome de Down. Daño y nexo causal. *Cuadernos Civitas de jurisprudencia civil*. 2008;76:279–290.
14. Julio César GC. Consejo genético. *Enciclopedia de Bioética y Bioderecho*. Spain: Comares Editorial; 2010.
15. Álvaro AGM. La regulación de los datos genéticos y las bases de datos de ADN. *Criterio Jurídico*. 2011;8(2):223–244.
16. Andrea MM, *Diagnóstico genético preimplantacional y responsabilidad médica por falsos negativos*. Madrid, Spain: REUS; 2018.
17. Morillo AM. El tratamiento de las acciones de wrongful birth y wrongful life a la luz de la nueva ley sobre interrupción voluntaria del embarazo. *Revista Jurídica de la Universidad Autónoma de Madrid*. 2011:23.
18. Casals MM, Fliu JS. 1966 SENTENCIA DE 18 DE MAYO DE 2006: Responsabilidad sanitaria derivada del nacimiento de un hijo con síndrome de Down. El médico no informó a la paciente del resultado negativo de la amniocentesis y de la necesidad de repetir la p ose rueba. Wrongful birth y wrongful life. *Cuadernos Civitas De Jurisprudencia Civil*. 2007;73:517–544
19. González MS .El tratamiento jurisprudencial del daño en las acciones de responsabilidad por wrongful birth. *Revista jurídica Universidad Autónoma de Madrid*. 2018:37