The Portuguese Prime Minister, *Pedro Miguel de Santana Lopes.*

Law n. º 12/2005 of 26 January

Personal genetic information and health information

The Assembly of the Republic decrees, in accordance with sub-paragraph c) of Article 161 of the Portuguese Constitution, the following law:

Article 1.

Object

The present law defines the concept of health information and of genetic information, the circulation of information and the intervention on the human genome on the health care system, as well as the rules for the collection and conservation of biological products for the purposes of conducting genetic tests or research.

Article 2.

Health information

For the purposes of the present law, health information is any information directly or indirectly linked to the present or future health status of a person, either living or deceased, including the person's clinical and family history.

Article 3.

Ownership of the health information

1. Health information, including registered clinical data, results of analysis and other subsidiary exams, interventions and diagnostics, is property of the person to whom it pertains; the health care system units are the custodians of such information and it cannot be used for any other purposes than health care and health related research, or other purposes defined by law.
2. The person to whom the health information pertains is entitled to take cognizance of his/her complete medical records, except in case of duly justified exceptional circumstances in which it is clearly demonstrated that the knowledge in question can be harmful or detrimental; he/she is also entitled to have his/her complete medical records made available to whomever he/she sees fit.
3. The access to the health information by the person to whom it pertains, or by a third party with consent, is made through an authorized physician chosen by the owner of the information.

Article 4.

Processing of health information

1. The persons responsible for the processing of health information must take all adequate measures to protect confidentiality, to ensure the security of the premises and the equipments and the control of the access to the information, as well as to guarantee the enforcement of the duty of confidentiality and of the rules of ethics and professional conduct by all health care professionals.
2. Health care system units must prevent undue access of third parties to all medical records and computer systems which contain health information, including access to its backup copies; they must ensure adequate levels of security and comply with all legal requirements regarding the protection of personal data, namely in order to avoid its accidental or unlawful destruction, its change, disclosure or unauthorized access as well as any other form of unlawful processing of the information.
3. Health information can only be used by the health care system subject to the conditions expressed on a written authorization from the person to whom it pertains or his/her representative.
4. Access to health information for research purposes can be given, as long as the information is anonymised.
5. Management of the systems that organize health information must guarantee the separation of health and genetic information from the remaining personal information, namely through the establishment of different levels of access.
6. Management of the information systems must guarantee the regular and frequent processing of backup copies of the health information, provided the guarantee of confidentiality established by law is ensured.

Article 5.

Medical information

1. For the purposes of the present law, medical information is the health information used for health care interventions or medical treatments.
2. “Medical records” means any kind of records, computerized or not, that contain health information about a patient or a member of his/her family.
3. Each medical record must contain all the available medical information about a patient, subject to the restrictions provided for in the following article.
4. The medical information must be recorded on the medical records by the physician that treated the patient or it must be computerised, under supervision of that physician, by another professional who is also subject to the duty of confidentiality, within the scope of competences specific of each profession and in the respect of its rules of ethics and professional conduct.

5. The medical records can only be consulted by the physician responsible for imparting health care treatments to the patient in question or, under supervision of the former, by another health care professional who is also subject to the duty of confidentiality and only to the extent needed to impart said treatments, without prejudice of epidemiological, clinical or genetic research that can be done about them, subject to what is defined in article 16.

Article 6.

Genetic information

1. Genetic information is the health information that deals with the hereditary characteristics of one or more persons, who are related or who have common characteristics of the genetic type, excluding, for the purposes of the present definition, the information obtained through blood relation tests or zygosity studies in twins, through studies of genetic identification for criminal purposes as well as the study of somatic genetic mutations in cancer.
2. Genetic information can result from genetic tests carried out by means of molecular biology, but also by means of cytogenetic, biochemical, physiological or imagiological tests, or through the simple collection of family information, registered as a family tree or in any other form, each of these tests being capable of expressing, by itself, the genetic status of a person or his/her family members.
3. Genetic information is considered medical information only when used for health care or health treatment purposes, for the confirmation or exclusion of a clinical diagnosis, in prenatal or preimplantation diagnosis, or for pharmacogenetics purposes; therefore, the information obtained through predictive tests used to determine de predisposition to common diseases and the presymptomatic tests for monogenic diseases is excluded.
4. Genetic information that does not have an immediate bearing on the patient's current status of health – like the information from paternity tests, from zygosity studies in twins and from predictive tests (with the exception of genetic testing to determine the response to drugs), as well as the information from heterozygosity, presymptomatic, prenatal and preimplantation tests – cannot be registered in the medical records, except in the case of genetic services or consultations that keep private separate files.
5. In case the medical records from genetic services or consultations contain genetic information regarding healthy persons, they cannot be accessed, shown or consulted by physicians, other health care professionals or staff of other departments of the health institution in question or of other institutions of the health care system.
6. Genetic information must be subject to legislative and administrative measures of reinforced protection in terms of access, security and confidentiality.
7. The use of genetic information is to be decided between the patient and his/her physician and it is subject to the rules of ethics, professional conduct and confidentiality that are binding to physicians and other health care professionals.

8. The existence of a working link or other type of relation between a physician or other health care professional and any other sector of activity – including insurance companies, professional entities or suppliers of goods and services of any kind – does not constitute a justification to the failure to comply with the obligation of confidentiality that binds the physicians and all other health care professionals.
9. Every citizen is entitled to know if a clinical record, a medical file or a medical or research record contains genetic information about himself/herself and his/her family, and is also entitled to know the objectives and the uses of the information in question, as well as the form in which it is stored and the period of time during which the information will be kept before destruction.

Article 7.

Genetic databases

1. "Genetic databases" means any kind of records, computerized or not, that contain genetic information about a group of persons or families.
2. The rules of creation, maintenance, management and securing of genetic databases for rendering health care services and relating to the research on health are regulated in accordance with the law regulating the protection of personal data.
3. The genetic databases that contain family information and the genetic records that allow the identification of members of a family must be managed and supervised by a medical geneticist or, if that is not feasible, by another physician.
4. Under the law, anybody can ask for and have access to personal information that is contained in personal data files.

Article 8.

Genic therapy

1. Medical interventions that have the purpose of intentionally modifying the human genome can only be carried out for preventive or therapeutic reasons and after verification of the conditions established by the present law.
2. All medical interventions that have the purpose of genetically manipulating characteristics that are considered normal – as well as the alteration of a person's germinal line – are strictly forbidden.

Article 9.

Genetic tests

1. Genetic diagnostic or pharmacogenetic testing must follow the general principles that regulate all other health care interventions or services.

2. The detection of the heterozygosity status for the diagnosis of recessive diseases, the presymptomatic diagnosis of monogenic diseases and the tests of genetic susceptibility in healthy persons can only be carried out by request of a medical geneticist, following a genetic counselling consultation and subject to the express written and informed consent of the person in question.
3. The results of genetic testing should only be communicated to the person whom they concern or, in case of diagnostic testing, to the legal representative or to the person designated by the person concerned and during a proper medical consultation.
4. In the case of heterozygosity, presymptomatic and predictive testing, the results should only be communicated to the person whom they concern and can never be communicated to third parties without his/her written express consent – this also refers to physicians or other health care professionals working for other departments of the health institution in question or for the same departments or services, but who are not involved in the process of testing of the person in question or of his/her family.
5. In case of prenatal and preimplantation testing, the results should only be communicated to the mother, to the parents or to their legal representatives.
6. Presymptomatic, predictive and preimplantation tests should not be performed in persons suffering from mental disablement and who cannot fully appreciate the implications of this type of tests or give their informed consent to its execution.
7. In situations of risk of severe, late-onset diseases that appear in the beginning of adulthood and that have no cure or proven effective treatment, the performance of any presymptomatic or predictive testing must be preceded by a previous psychological and social evaluation and by the follow-up of the patient after the delivery of the tests results.
8. The frequency of the genetic counselling consultations and the form of the psychological and social follow-up should be determined based on the severity of the disease, on the usual age of manifestation of the first symptoms and on the existence, or not, of a proven treatment.

Article 10.

Heterozygosity, presymptomatic, predictive and prenatal testing

1. For the purposes of the previous article, the tests used for the detection of heterozygosity status are those that allow detecting healthy persons who are heterozygosity gene carriers for recessive diseases.
2. Presymptomatic tests are those that allow detecting persons who are still asymptomatic but carry the genotype that is unequivocally responsible for a monogenic disease.
3. Genetic predictive tests are those that allow detecting susceptibility genes – this means a genetic predisposition for a particular disease that has complex heredity and usually begins in adulthood with a late onset.

4. Pharmacogenetic tests are the predictive tests that allow detecting a predisposition to differential responses in a treatment with a specific drug or the susceptibility to adverse reactions derived from the toxicity of the drug in question.
5. Prenatal tests are all the tests performed before or during pregnancy, with the objective of obtaining genetic information about the embryo or the foetus; the preimplantation diagnosis is considered a particular case among this type of tests.
6. Screening tests are all diagnostic tests and heterozygosity, presymptomatic, predictive and prenatal tests that are performed on the entire population or on population groups that have an increased risk of contracting a specific disease, namely due to gender, age or ethnic origin, in any time in life.

Article 11.

Principle of non-discrimination

1. No person can be prejudiced, in any way, due to the presence of a genetic disease or due to his/her genetic heritage.
2. No person can be discriminated, in any way, due to the results of a genetic diagnostic test or due to heterozygosity, presymptomatic, predictive or prenatal tests, including those performed with the purpose of obtaining or maintaining a job, subscribing health and life insurances, having access to education, as well as for purposes of adoption, whether regarding the adopter or the adoptee.
3. No person can be discriminated, in any way, in what concerns his/her right to obtain medical, psychological and social follow-up as well as genetic counselling because he/she refused to be subject to a genetic test.
4. A fair and equitable access to genetic counselling and genetic testing is guaranteed to all persons; however, the needs of the populations that are more strongly affected by a specific genetic disease or diseases are duly safeguarded.

Article 12.

Genetic tests and insurance policies

1. Insurance companies cannot ask for a genetic test or use any kind of genetic information already available as a means to refuse the subscription of a life or health insurance or to establish a higher premium.
2. Insurance companies cannot ask their clients to submit to genetic tests for the purposes of subscribing a life or health insurance or for any other purposes.
3. Insurance companies cannot use genetic information obtained through genetic tests performed previously on their clients, whether actual or potential, for the purposes of subscribing a life or health insurance or for any other purposes.

4. Insurance companies cannot demand or use genetic information obtained through the collection and registration of the client's family history as a means to refuse the subscription of an insurance, to establish a higher premium or for any other purposes.

Article 13.

Genetic tests on the workplace

1. The employment of new workers cannot depend on a selection process based on the demand or the performance of genetic tests or on the results of previous genetic tests.
2. Companies and other employers are not allowed to demand that their employees submit to genetic tests or disclose the results of previous genetic tests, even with the employees consent.
3. In case of hazardous working environments, where there is a risk of specific hazards for workers who have a particular disease or susceptibility, or where there is a risk of the environment affecting the workers' ability to safely perform a particular task, the relevant genetic information can be used for the benefit of the worker – but never to his/her prejudice – provided the purpose of that information is to protect the person's health, his/her personal security and that of the other workers, provided the test is performed after obtaining the worker's informed consent and following appropriate genetic counselling, provided the results are disclosed only to the person concerned and also provided that the worker's employment situation is never put at risk.
4. The specific situations that imply serious risks to public health and safety can constitute an exception to what was stipulated on the previous paragraphs, provided that the restriction imposed by the next paragraph is adhered to.
5. In case of the situations explained on the previous paragraphs, the genetic tests – regarding exclusively a serious risk to the worker's health or relevant to his/her current health status – must be determined, performed and supervised by an independent agency or entity and never by the employer.
6. The employer is responsible for paying the expenses arising from the genetic tests performed on demand of the employer or on his direct interest.

Article 14.

Genetic tests and adoption

1. No genetic testing or any kind of previously available genetic information can be requested or used for the purposes of adoption.
2. The adoption services or the prospective parents cannot request genetic tests or use any information from previous genetic tests done on the adoptees.
3. The adoption services cannot demand that the adopter parents submit to genetic tests nor use any genetic information previously available regarding the adopters.

Article 15.

Laboratories that perform or provide genetic tests

1. The government must regulate the conditions relating to the offer and the performance of genetic tests of heterozygosity status, presymptomatic, predictive or prenatal and preimplantation tests, in order to avoid that these tests are made by national or foreign laboratories that do not have the support of a proper and multidisciplinary medical team, and to avoid the possible over-the-counter marketing of this type of tests.
2. Under the law and subject to the ethical, quality and safety recommendations from national and international regulatory bodies, the Government is responsible for determining the accreditation and certification measures with which the public and private laboratories that perform genetic tests must comply; the Government is also responsible for licensing such laboratories.

Article 16.

Research on the human genome

1. Research on the human genome must follow the general rules of scientific research related to health, and its researchers have an additional duty of confidentiality regarding the identity and the characteristics of the persons studied individually.
2. The scientific community must have free access to the data resulting from research on the human genome.
3. Research on the human genome is subject to the approval by ethic commissions from hospitals, universities or research institutions.
4. The research on the human genome on individuals cannot be done without previously obtaining an informed written consent from the persons in question, given after the explanation of their rights, of the nature and purposes of the research, of the procedures used and of the potential risks for the persons involved and for others.

Article 17.

Duty of protection

1. It is illicit to create any list of diseases or genetic characteristics that may support the request of diagnostic tests, of heterozygosity, presymptomatic, predictive or prenatal tests or of any king of genetic screening.
2. Every citizen has the right to refuse submitting to a genetic test concerning the heterozygosity status or to a presymptomatic, predictive or prenatal test.
3. Every citizen has the right to receive genetic counselling and, if appropriate, psychological and social support, before and after submitting to heterozygosity, presymptomatic, predictive or prenatal tests.

4. The genetic testing of minors can only be requested if the tests are done in the benefit of the minors – and never in their prejudice – with written consent from their parents or legal tutors and always seeking first the minors own consent.
5. In particular, no predictive tests on minors can be requested, regarding late-onset diseases that usually begin in adulthood and that have no cure or proven effective preventive treatment.
6. The former also applies to the prenatal diagnosis of diseases that usually begin in adulthood and that have no cure – the diagnosis cannot be done just for information of the parents but only with the aim of preventing the disease or the deficiency and this within the time limits established by law.
7. Physicians have the duty of informing the persons who consult them about the transmission mechanisms and about the risks that they imply for their relatives, as well as guiding the persons in question to a medical genetic consultation, which must be ensured in accordance with the regulatory legislation of the present law.
8. In case of genetic screening of the population, in addition to the individual rights, the rights of the population or groups of the population must also be protected, in order to avoid their stigmatization.
9. The persons who have special needs, as well as those who have an impairment or suffer from a chronic illness, including patients with genetic pathologies and their families, are entitled to the State's protection in regards to information about the health care they need.

Article 18.

Collection and conservation of biological material

1. The collection of blood and other biological products, as well as the collection of DNA samples for genetic testing, is subject to separate informed consents for health care purposes and for biomedical research purposes; the consent must include the purpose of the collection and the duration of storage of the samples and its by-products.
2. The stored material remains property of the person from whom it was collected and, in case of death or incapacity of the person in question, of his/her relatives.
3. The person to whom the biological material belongs can withdraw, at any time, his/her consent or, in case of death or incapacity of the person in question, his/her relatives can withdraw it; if that is the case, the stored biological samples and its by-products must be destructed for good.
4. The biological samples collected for a particular purpose should not be used for other health care or biomedical research purposes unless the person from whom it was collected gives consent to it or, in case of death or incapacity of the person in question, his/her relatives give consent or after irreversible anonymization.

5. The samples that are collected for a specific medical or scientific purpose can only be used with express consent from the persons involved or from their legal representatives.
6. In special circumstances, in which the information can be important to enable the treatment or the prevention of a genetic disease in the family, that information can be processed and used in the context of genetic counselling – even if it is no longer possible to obtain the informed consent from the person to whom it belongs.
7. All relatives in direct line of ascent or descent, as well as second degree relatives, can have access to a stored sample of genetic material, provided that it is necessary to obtain a better knowledge of their own genetic status, but not to know the genetic status of the person to whom the sample pertains or of other family members.
8. The commercial use, the patent registration or any type of financial gains derived from biological samples, as such, is strictly forbidden.

Article 19.

DNA and other biological material databanks

1. For the purposes of the present law, “biological material databanks” are defined as any collection of biological samples or its by-products, with or without a storage time limit, previously accumulated or prospectively performed, obtained through routine health care provision, whether in screening programs or for research purposes, and that includes samples that are identified, identifiable, anonymised or anonym.
2. No person can collect or use human biological samples that have already been collected, or its by-products, with the purpose of creating a biological material databank, without first obtaining authorization from an entity accredited by the health authorities and by the National Data Protection Commission if the biobank is associated to personal information.
3. Biological material databanks should only be created for the purposes of providing health care services, including the diagnostic and the prevention of diseases, or for the purposes of basic or health related research.
4. Biological material databanks should only accept samples in response to requests from physicians and not from the persons in question or their relatives.
5. Written informed consent is necessary to obtain and use material for a biological material databank; the consent form should include information about the purposes of the databank, the person responsible for it, the types of research it performs, its potential risks and benefits, the conditions and duration of storage, the measures taken to guarantee the privacy and confidentiality of the persons involved, as well as the prevision regarding the possible disclosure or not of the results obtained from the materials in question.
6. In case of retrospective use of the samples, or in special situations in which it is not possible to obtain consent from the persons involved – due to the amount of data or of subjects, to

their age or other similar reason – the material and the data can only be processed for the purposes of scientific research or for collecting epidemiological or statistical data.

7. The conservation of samples of dried blood on paper, obtained in neonatal screenings or others, must be considered in the light of the potential benefits and risks it poses to individuals and to the society; however, those collections can be used for family studies in the context of genetic counselling or for genetic research, provided they are previously anonymised in an irreversible way.
8. The privacy and confidentiality must always be ensured – the storage of identified material should be avoided, the access to the collections of biological material should be controlled, the number of persons authorized to access it should be restricted and its safety should be guaranteed, particularly in terms of losses, changes and destruction.
9. Only anonym or irreversibly anonymised samples can be used, the use of identified or identifiable samples should be limited to studies that cannot be conducted in any other way.
10. Commercial entities cannot store or use human biological material that has not been anonymised.
11. If there is an absolute need to use identified or identifiable samples, these should be coded and the identifying codes must be kept separately, but always in a public institution.
12. If the databank has identified or identifiable samples and if the communication of the results of studies performed can be foreseen, a medical geneticist should be involved in the process.
13. The stored biological material is considered to be the property of the person from whom it was collected and, in case of death or incapacity of the person in question, of his/her family members; it should be stored for as long as it proves to be of use for present and future relatives.
14. The researchers responsible for the studies on samples stored in biological material databanks should always check that the rights and interests of the persons to whom the biological material pertains are protected in the context of the genetic tests performed on those persons or on their relatives; this includes the protection of their privacy and confidentiality, but also the preservation of the samples, that may be necessary to diagnose family diseases in the future.
15. It is the researchers responsible for the collection and maintenance of biological material databanks duty to watch over their conservation and integrity and to inform the person from whom consent was obtained of any loss, change or destruction, as well as of the decision to abandon the research in question or to close the databank.
16. The law defines the rules for the licensing and the promotion of quality assurance processes of biological material databanks.
17. The transfer of a large number of samples or of biological material collections to other national or foreign entities must always respect the purposes for which the bank was

originally created and for which consent was obtained, and must also be approved by the responsible ethic commissions.

18. The creation of databanks that describe a particular population and the possible transfer of its data must be approved by the National Council of Ethics for Life Sciences; if the databanks are representative of the national population they must be approved by the Assembly of the Republic.
19. The biological material databanks created for forensic purposes, criminal identification purposes or others must be the object of a specific regulation.

Article 20.

Human genetic heritage

Human genetic heritage is not susceptible of any patent registration.

Article 21.

Report on the application of the law

The Government, having heard the opinion of the National Council of Ethics for Life Sciences, shall present to the Assembly of the Republic, two years after the coming into force of the present law and every two years thereafter, a report that presents an inventory of the conditions and the consequences of the application of the law, having regard to the public debate about its ethical principals and the scientific progress that was achieved.

Article 22.

Regulation

1. The Government has 180 days to regulate the present law.
2. The definition of measures of promoting the research and protection of personal genetic identity, of clinical and analytical validation of genetic tests, in particular of predictive tests to detect susceptibility genes and the response to drug treatments, as well as the genetic screening tests, will be the object of a separate regulation.

Approved on December 9 2004.

The President of the Assembly of the Republic

João Bosco Mota Amaral.

Promulgated on January 7 2005.

To be published.

The President of the Republic, JORGE SAMPAIO.

Approved by referendum on January 13 2005.

The Portuguese Prime-Minister, *Pedro Miguel de Santana Lopes.*