Portuguese Law 12/2005 (26 February 2005)

Defines health information as any information directly or indirectly linked to the present or future health status of a person, either living or deceased, including clinical and family history

Health information is property of the person on whom it pertains (though access to it is made through an authorized physician) and cannot be used for any other aims than health care and research, or other defined by law

Defines medical information as the health information used for medical intervention

Defines genetic information as the health information linked to genetic characteristics of one or more related persons (excluding, for the purposes of the law, identity and forensic testing, as well as somatic mutations), obtained through any means, including molecular genetic, cytogenetic, biochemical, physiological tests or imagiology, and pedigree information

Genetic information is considered to be medical information only when used for the confirmation or exclusion of a clinical diagnosis, in prenatal or preimplantation diagnosis or for pharmacogenetics purposes, excluding presymptomatic or susceptibility testing

Only information with immediate interest for the patient's current status of health (diagnostic and pharmacogenetic information) can be entered in general hospital records; information from presymptomatic, susceptibility, prenatal, preimplantation forensic and identity testing can only be registered in records of genetic services that keep separate files (and these cannot be accessed by other professionals of the same or of other health institutions)

Defines a genetic database as any register, either in an informatics support or not, containing genetic information on persons or families; if a database or a genetic registry includes any kind of family information it should be curated by a medical geneticist

Diagnostic or pharmacogenetic testing should follow the general principles of all other health care intervention

Carrier, presymptomatic and susceptibility testing should be preceded by genetic counselling and written informed consent, and requested through a medical geneticist

Presymptomatic, susceptibility and preimplantation diagnosis should only be performed in persons that can fully appreciate all their implications and give their consent

In case of risk for a severe, late-onset disease that has no effective treatment, any predictive testing should be preceded by a psychosocial evaluation and followed after result delivery

Insurance companies cannot ask for a genetic test or use any kind of genetic information already available (including pedigree information) to refuse life or health insurance or establish a higher premium

Employers cannot ask for or use any kind of genetic information, even with the workers' consent, except for their health protection (in case of hazardous environments), and only if done in the context of genetic counselling and if their employment is not put at risk; the exception could be made in case of serious risk to public security or public health, in which case genetic testing should be conducted by an independent entity

No genetic testing or any kind of genetic information can be requested in case of adoption, both to the adoptees or the prospective parents

In the case of minors, genetic testing should be done only in their benefit, after written consent from their parents or legal tutors, but also procuring the minors consent

Nevertheless, in the case of severe and untreatable diseases, with onset usually in adult life, predictive testing cannot be performed in minors; and prenatal testing should not be done just for information of the parents but only with the aim to prevent the birth of an affected child (termination of pregnancy is legal for genetic reasons within the first 24 weeks, and up to term in case of early lethality, e.g., anencephaly)

The government must now regulate the offer of genetic testing, in order to avoid its direct marketing to the public or by public or private laboratories, outside of the context of genetic counselling

In case of population screening, the rights of the population or groups of the population should also be protected, in addition to the individual rights

Collection, conservation and usage of biological samples for genetic testing should be subject to an informed consent separate for health care and biomedical research, including its purposes and duration of storage

If consent for a different purpose cannot be obtained, e.g. in case of death, stored samples can be used in the context of genetic counselling, in order to enable treatment or the prevention of a genetic disease in a relative (but not to know the genetic status of other family members)

Biological samples cannot be used for any commercial purposes; commercial entities cannot store or use identified or identifiable samples; if absolutely needed, coded samples can be used, if the identifying codes are kept in a public institution

A biobank is defined as any collection of biological samples or its derivatives, previously accumulated or prospectively performed, obtained through health care provision, population screening or research, with or without any identification, and with or without a time limit

Previous authorization must be requested from the health authorities and, in case of identified or identifiable samples, from the national personal data protection agency

A biobank must have a health care or a (basic or applied) health research purpose; if communication of results can be foreseen, a medical geneticist should be involved

When consent is not possible (proband deceased) or easily obtainable (e.g. large amount of samples needed), samples can be used for family studies, or can be processed for epidemiological or statistical purposes, only if previously and irreversibly anonimised.