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POLICY

Council of Europe adopts protocol on genetic testing for health purposes

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A new Additional Protocol to the Convention on Human Rights and Biomedicine

On 7 May 2008, the first international legally binding instrument concerning genetic testing for health purposes was adopted by the Committee of Ministers of the Council of Europe. (The Council of Europe, set up in 1949, is an intergovernmental organization with a pan-European vocation that aims at fostering a closer cooperation between its 47 Member States (which include the 27 EU members) for the protection of human rights and the promotion of democracy. Since the beginning of the 1980s, this organization has been active in the field of bioethics. A substantial legal corpus has been developed laying down fundamental principles. The reference instrument is the Convention on Human Rights and Biomedicine (ETS no. 164, 1997).)

The sequencing of the human genome and the development of new technologies make human genetics a very dynamic sector. The very rapid progress in this field has prompted the Council of Europe to focus on the ethical and legal issues raised by applications of genetics, in particular genetic testing, and to draw up legal standards to protect fundamental human rights with regard to these applications.

Protection and guarantees in the fields of biology and medicine, including human genetics, are provided by the Council of Europe Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Applications of Biology and Medicine (Convention on Human Rights and Biomedicine). (Convention on Human Rights and Biomedicine (Oviedo Convention) (ETS no. 164)). This Council of Europe convention was opened for signature on 4 April 1997 and has been signed by most European states. It sets out fundamental principles applicable to routine medicine and those that apply to new technology in the area of human biology and medicine. It also serves as a reference instrument for the European Union and for other international organizations, such as UNESCO and WHO. The Convention sets out a number of principles with regard to genetics (Articles 11–14), particularly genetic testing and interventions on the human genome.

To develop and supplement the principles set forth in the Convention, the Council of Europe Steering Committee on Bioethics (CDBI) elaborated a new Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes. This Protocol was adopted by the Committee of Ministers of the 47 Member States of the Council of Europe on 7 May 2008 and opened for signature on 28 November 2008.

Embryos and fetuses excluded

The Protocol covers all genetic testing carried out for health purposes, except those with regard to the human embryo and fetus and to that carried out for research purposes, which are covered by another Additional

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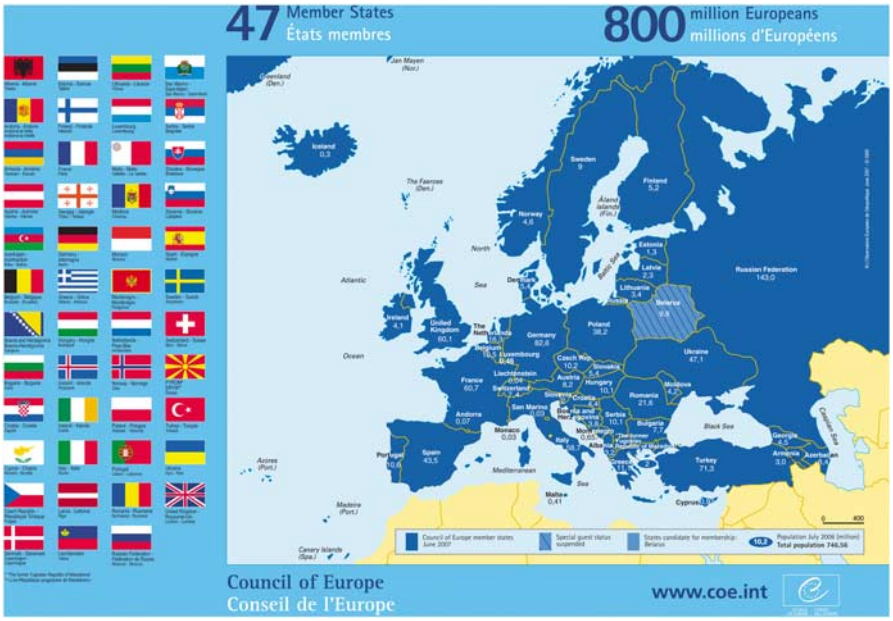

Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research.

When deciding to focus on genetic testing for health purposes, the Steering Committee on Bioethics (CDBI), which elaborated the draft Protocol, was guided by practical consideration with regard to a specific medical field involving different categories of professionals, which will be concerned by the provisions of the Protocol.

Genetic test: chromosome, DNA, RNA or relevant biological information

The Protocol applies to genetic tests, which are carried out for health purposes, involving analysis of biological samples of human origin and aiming specifically to identify the genetic characteristics of a person that are inherited or acquired during the early development (Article 2.1). For the purpose of the Protocol, the notion of analysis refers to chromosomal analysis, DNA or RNA analysis or analysis of any other element enabling information to be obtained which is equivalent to that obtained using the first two methods (ie, information directly linked to the genetic characteristics sought) (Article 2.3.a).

The Protocol lays down principles with regard to, in particular, the quality of genetic services, prior information and consent as well as genetic counseling. A whole chapter is dedicated to the regulation of genetic testing on persons not able to consent; the issue of genetic testing for the benefit of family members is specifically addressed. The Protocol covers the protection of private life and the right to information obtained by means of genetic testing. It also addresses the issue of genetic screening.

The field of genetics is technically complex and constantly evolving. In addressing the issue of genetic testing for health purposes, the CDBI was concerned that the technical terms corresponded to the generally accepted standards at the international level. To this end, it consulted different experts in genetics and used as a basis the recommendations of the European Society of Human Genetics and also took into account the OECD guidelines on quality assurance in molecular genetic testing.

Good medical practice requires clinical utility

The ESHG recommendations were particularly important for the elaboration of provisions with regard to genetic services. In Article 5 on the quality of genetic services, the provisions relate to genetic tests, which shall meet generally accepted criteria of *scientific validity and clinical validity*, as well as laboratories and persons providing genetic services.

Particular emphasis is also placed on the importance of taking into account the *clinical utility* of a genetic test as an essential criterion for deciding to offer this test to a person or a group of persons. Taking it into account can be regarded as an integral part of good medical practice with regard to any decision to carry out a test. This was considered to be of particular importance for tests proposed outside any individualized medical supervision. The Explanatory Report to the Protocol (paragraph 62) states that existing evidence on a genetic test's clinical utility has thus to be available, in particular to the health professionals and to the persons concerned by the test.

Such evidence should be obtainable from the laboratory carrying out the test.

Two issues were particularly discussed during the elaboration process of the Protocol: information and genetic counseling and tests directly accessible.

Genetic counseling needed: 'light' or 'heavy and long'

The Convention on Human Rights and Biomedicine requires in its Article 12 that

> 'tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counseling.'

These requirements are reiterated in Article 8 of the Additional Protocol. The expression 'appropriate genetic counseling' is not defined in the Convention, but its scope has been further discussed in the framework of the elaboration of the Additional Protocol. The Explanatory Report to Article 8 of the Additional Protocol specifies that genetic counseling is an individualized process taking into account, in particular, the psychological and family context of the person concerned and involving an exchange between him or her and the person providing the counseling. It was therefore considered that genetic counseling could vary in form and extent in accordance, in particular, with the implications of the test and their significance for the person concerned or the members of his or her family, including possible implications for procreation choices. Genetic counseling could thus go from being a 'very heavy and long' procedure to a 'lighter' one, which may be limited to a pre-test counseling; hence the adjective 'appropriate.'

Medical supervision to guarantee quality of information and support

The quality of the prior information given to persons envisaging genetic testing, designed to enable them to take an informed decision, as well as the support provided to persons taking such decisions and dealing with the implications of a test and its results, were two main concerns for the CDBI in its work on genetic testing. The Committee took the view that such testing should be carried out only in response to a specific indication made on the basis of a precise evaluation, by a doctor, of the situation of the person concerned. The Protocol thus lays down (Article 7.1) the general rule that genetic testing for health purposes may be carried out only under individualized medical supervision.

Test directly accessible

In this context, the CDBI examined specifically the issue of tests that are directly available to the individuals, ie, outside any conventional medical system.

The Protocol prohibits exceptions to the general rule defined in Article 7.1 in the case of genetic testing with important implications for the health of the person concerned or his or her family members or for procreation choices.

The key concerns here are the proper interpretation of predictive test results and the guarantee of an appropriate genetic counseling to understand its implications. The results of such genetic tests may be particularly complex to interpret and may, for instance, require that additional medical information or information about family history be taken into account. In the case of many predictive tests, even though the test may reveal a high probability of developing a particularly serious disease, the time of onset of the disease and the severity of the symptoms are often uncertain. Finally, the problem of understanding the nature of the test and the implications, including the implications for family members, the potential psychological impact of the results on the person concerned and the often important decisions facing that person require that such tests be carried out under individualized medical supervision.

No serious consequences, more freedom

The Protocol does, however, provide that states under certain conditions may make exceptions to the general rule in the case of tests that would not have serious implications, the principal aim being to ensure compliance with the provisions of the Protocol with regard to the nature and quality of the prior information, free and informed consent and genetic counseling (Article 7.2).

Each state therefore has a degree of discretion in the decision to allow a test to be carried out without individualized medical supervision, and when it comes to the procedures to be followed and the bodies involved in this process.

Protect against direct to consumer testing where appropriate

However, as the objective is to protect the person concerned, particular account must be taken of the importance of the potential implications of the test in question for the person on whom it is to be carried out or on his or her family members, the ease of interpretation of

the results and, where appropriate, the treatment possibilities for the disease or disorder concerned.

On that basis, the Council of Europe will continue to follow the evolution of the offer of directly accessible tests, with a view to facilitating the implementation of the principles laid down in the Protocol.

Do good, do not harm with genetics

The preamble of the Additional Protocol underlines the role of genetics in contributing to the progress in biomedical sciences in reducing morbidity and mortality and in improving the quality of life. The objective of the Protocol is to protect against the improper use of genetic testing, by providing a framework for the protection of fundamental rights and freedom with regard to such a test. Scientific developments will continue to be followed and taken into account in the re-examination process foreseen by Article 23 within 5 years of the entry into force of the Additional Protocol with a view to ensuring the proper protection of individuals and promoting thereby the benefits of genetic testing for their health.

After genetic testing for health purposes, the CDBI is now considering the issue of predictivity and genetic testing in the field of insurance with a view to the elaboration of a new legal instrument.

Further reading

The text of the Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes as well as its Explanatory Memorandum can be consulted at http://conventions.coe.int/Treaty/EN/Treaties/Html/203.htm.

Further information on the work of the Council of Europe as well as on adopted legal instruments in the field of bioethics can be found at http://www.coe.int/bioethics.