Abstract

A European Commission (EC) Expert Group emphasized, in 2004, that “a consensus definition of genetic testing should be developed globally”. The need for an agreement of a common definition was also identified at an EC informal network on genetic testing by representatives of the Member States of the European Union. The purpose of this work was the collection and analysis of European and other legislation on genetic testing and comparison of the definitions therewith contained. Documents were analysed for type and object of testing, as used in the 1st background document on Definitions of Genetic Testing, and compared by legal fields (insurance and labour law, privacy and confidentiality, data protection, biobanks, health care and forensics); some (often supranational) laws are very complex and dealt with various legal fields at the same time. There was no standard for the definitions used in the legal documents analysed, but different approaches were identified (some used a more general, while others applied a more specific definition of genetic testing). Since definitions are core elements of legal documents, their harmonization within a particular legal field is very needed, but will also be very challenging.
INTRODUCTION

A European Commission (EC) Expert Group produced the document “25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing, 2004”\(^1\). In this document, the working group established, as its very first recommendation, the need to “refer precisely to an explicit definition of the terms used or topic addressed” on any official statement or position, but also the need to develop “a consensus definition of genetic testing should be developed globally”\(^1\).

That document suggested that this definition should be developed by “all respective public and private bodies involved (including the World Health Organisation, the Organisation for Economic Co-operation and Development, the European Commission, the International Federation of Genetic Societies, and the International Conference on Harmonisation)” ; and that “the European Commission should consider taking the initiative” on the topic.

In this document, the expert group used a broad definition for genetic testing, i.e., “*any test that yields genetic data*\(^2\), unambiguously revealing underlying DNA information, either germ-line or somatic and where the nature/technology of the test is not important, but the information derived."

The need for an agreement of a common definition was also identified at the meeting of an informal network with Member-States (MS) representatives, which was a follow up of the expert group.

As a consequence, the EC asked the European NoE EuroGenetest\(^2\) to prepare such a definition. This was preceded by a review of definitions of genetic testing in documents (recommendations, guidelines, other) from several of the main international and European Institutions, but also from professional organizations (genetics and other), official government regulatory bodies and agencies, national health institutions, pharmaceutical industry, insurers, ethical organizations, patient associations, human rights organizations, among others.

With the present document, we extend now that analysis and reflection to the definitions of genetic testing contained in European and other legal documents.

AIMS

The major general aim of this work was to contribute to the discussion of a consensus definition and of its global applicability.

Among the specific aims of WP3.4 were the comparison of definitions of genetic testing contained in legal documents from official bodies in Europe and its Member-States (MS) and that from other countries (USA, Canada, Japan, Australia and others).

Another aim was to compare these legal documents regarding genetic testing, based upon those definitions, and the context in which they were used, and relate them to the specific objectives and purposes of that legislation.
METHODS

The expression “legal document” means all documents that were, directly or indirectly, created by an
authorised state (public) organisation: the legislator was either a governmental body or an advisory
board/council/working group nominated by it.

Legal documents at the supranational level, from European Organizations (EU) and Council of Europe
(CoE), already listed and analysed in the 1st background document on Definitions of Genetic Testing
v.3 were used here again, as was the recent “Additional Protocol” to the Oviedo Convention, to
provide a comparison with national laws.

For the purpose of this document, a law is defined here as any type of legal norm (act, regulation,
resolution, convention, etc.), independently from its binding or non-binding status. A law is binding
only if it can be enforced. This distinction, however, is rarely used in international law, as
international law is not based on bindingness.

Other (i.e., non-legal) non-binding norms and codes (e.g., professional and other guidelines and
recommendations) were thus not included here (they are reviewed and analysed in the first
background document).

Generally speaking, the role and status of the international law becomes close to that of national
(domestic) legislation, when it is incorporated into the national law in the proper way. While national
documents can be legally binding or non-binding, all the supranational legislation examined here was
non-binding. It should be noted, however, that a non-binding supranational document, as the Oviedo
Convention, becomes legally binding after ratification in a given country.

Given that only a few countries have legally-binding legislation regarding genetic testing, we decided
to extend the search and comparison to non-binding legislation as well, whenever enacted by an
official body (e.g., regulatory agencies and other linked in any way to a national government or to
European institutions).

This study was based on a search within various online resources. Several legal international
databases (specialized on genetics) and one general legal database were used systematically, to
collect national legislation concerning genetic testing, in addition to national ones.

Two of these belong to World Health Organization (WHO): (1) one is the Ethical, Legal, Social
Implications & Issues of Human Genome Project Genetics Resource Directory (ELSI ReD), which
aims “to provide ready access to existing legislation and policy documents relating to various aspects
of genetic research and applications”; (2) the second is the International Digest of Health Legislation
(IDHL), which contains a selection of national and international health legislation, relating to health
care, in every member state of WHO.

(3) The HumGen database is an international resource, concerning ethical, legal and social issues in
human genetics. (4) The Center for Ethics and Law in Biomedicine (CELAB) also established a
database on law, ethics and policy in the field of biomedical law including human genetics.

(5) The Organisation for Economic Co-operation and Development (OECD/OCDE) database, (6) the
EuroGentest database on Regulations and Practices Related to Genetic Counselling in 38 European
Countries and (7) the European Ethical-Legal Papers series are all based on surveys, and thus more refined and better analyzed than simple collections of data.

(8) A well-known general legal-database (world law guide) is Lexadin, which includes laws/rules on health care, insurance, labour, private law, etc., was also used.

(9) Finally, national legal databases were also searched, whenever possible (i.e., if there were no language or technical problems).

All these databases were searched in every country and for international legislation, using keywords such as “genetic testing” and/or “genetic test(s)”, as well as specific types of genetic tests and related synonyms (diagnostic genetic test/testing, confirmation/confirmatory test, genetic diagnosis, presymptomatic test/testing, predictive test/testing, disease predisposition test/testing, susceptibility test/testing, testing for complex diseases, drug response test/testing, pharmacogenetics test/testing, adverse reactions, dose adjustment, carrier test/testing, heterozygote/heterozygosity test/testing, prenatal test/testing, fetal testing, PGD, preimplantation/pre-implantation test/testing/diagnosis/genetic test/testing/diagnosis, population screening, population genetic screening, genetic screening, predictive test/testing, carrier testing, identity (genetic) testing, forensic testing, civil identification, criminal identification, paternity testing, family testing, twin/zigosity testing, chromosomes/chromosomal analysis, cytogenetic(s), genes, nucleic acid(s), genetic material, gene products, biochemical testing, genetic biochemistry, metabolic (products), protein, hormone, enzyme activity, RNA, metabolites, routine blood tests, clinical pathology, blood tests/exams/workup, family history/information/tree, pedigree (information), genealogy/genealogical, genogram). When these keywords gave no results for a given database, the words “genetic” and “DNA” were searched separately, as well.

These legal documents were all included in a database and analysed with the NVivo8 software (QSR International Pty Ltd, Doncaster Victoria, Australia), which allows the analysis of text data.

The documents were analysed and compared by legal fields, and for the same items (type of testing, object of test, etc.) as used in the 1st background document Definitions of Genetic Testing v.3. Hence, only the comparison based on the legal fields will be discussed below.

RESULTS

European Union (EU) institutions – European Commission (EC) and Parliament (EP) – and the Council of Europe (CoE), individual EU Member-States (MS) and other European countries, as well as a few non-European countries (Australia, Canada, Japan, USA) have all been surveyed for legal documents containing definitions of genetic testing, between March and October 2008. Table 1 shows the national legislation identified for all the countries considered, whether those legal norms were available or translated to English, and whether they contained definitions of genetic testing (or any type of genetic testing).
Table 1. Definitions of genetic testing and their documents

<table>
<thead>
<tr>
<th>EUROPEAN UNION (EU) INSTITUTIONS AND COUNCIL OF EUROPE (COE)</th>
<th>EUROPEAN UNION (EU27) MEMBER-STATES COUNTRIES</th>
<th>OTHER EUROPEAN COUNTRIES</th>
<th>NON-EUROPEAN COUNTRIES</th>
</tr>
</thead>
<tbody>
<tr>
<td>EC 25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing</td>
<td>Austria Austrian Gene Technology Act</td>
<td>Switzerland</td>
<td>Australia Genetic Testing - Guidelines for prioritising genetic tests</td>
</tr>
<tr>
<td>EC Ethical, Legal and Social Aspects of Genetic Testing: research, development and clinical applications, STRATA Group on Genetic Testing</td>
<td>Belgium Genetics and Insurance</td>
<td></td>
<td>Australia Ethical guidelines on the use of assisted reproductive technology in clinical practice and research (as revised in 2007 to take into account the changes in legislation)</td>
</tr>
<tr>
<td>EC Opinion on the Group of Advisers on the Ethical Implications of Biotechnology to the European Commission</td>
<td>Belgium Royal Decree</td>
<td></td>
<td>Australia ALRC 96 ‘Essentially Yours: The Protection of Human Genetic Information in Australia, This Report reflects the law as at 14 March 2003</td>
</tr>
<tr>
<td>EC Opinion on the ethical aspects of genetic testing in the workplace - Opinion N° 18</td>
<td>Bulgaria Bulgarian Health Act</td>
<td></td>
<td>Canada Genetic testing and privacy</td>
</tr>
<tr>
<td>EC Towards quality assurance and harmonisation of genetic testing services in the EU</td>
<td>Cyprus The Safeguarding and Protection of Patients’ rights Law, 2004</td>
<td></td>
<td>Canada CCHF Molecular Genetics Guidelines</td>
</tr>
<tr>
<td>CoE Convention for the Protection of Human Rights and Dignity of the Human Being (and it’s additional protocol for genetic testing)</td>
<td>Denmark Law on artificial fertilization in connection with medical treatment, diagnosis, and research (Research on embryonic stem cells).</td>
<td></td>
<td>United States Executive Order, To Prohibit Discrimination in Federal Employment Based on Genetic Information</td>
</tr>
<tr>
<td></td>
<td>Denmark Act on the use of health data etc. on the labour market</td>
<td></td>
<td>United States Genetic Information Non-Discrimination Act of 2007 (Placed on Calendar in Senate)</td>
</tr>
<tr>
<td></td>
<td>Estonia Human Genes Research Act</td>
<td></td>
<td>United States Genomics and Personalized Medicine Act of 2007 (Introduced in Senate)</td>
</tr>
<tr>
<td></td>
<td>Finland Act on the Protection of Privacy in Working Life</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
No relevant legislation regarding genetic testing could be found for Albania, Belarus, Bosnia-Herzegovina, Croatia, Czech Republic, Liechtenstein, Lithuania, Luxembourg, Macedonia, Moldova, Monaco, Poland, Romania, Russia, San Marino, Serbia, Slovenia, Turkey and Ukraine (Table 1). For some of the legal documents found from other countries (Austria, Belgium, France, Greece, Spain, Sweden), no definitions of genetic testing were found or no English translation was available, in spite of efforts to get at least a translation of the most relevant parts.

Most of the documents identified related to the following legal fields: (1) labour law, (2) insurance, (3) biobanks, (4) data protection, (5) health care or regulation of medical activities, (6) forensic/criminal/penal law, (7) family law, (8) general data protection or privacy/confidentiality law, and global (9) supranational laws.

The total number of legal documents identified was 67 (59 national from 48 countries and 8 international documents, from EU and Council of Europe). Their availability in English allowed the analysis of 60 of these: 52 were national and 8 were supranational legislation. The Appendix includes the weblinks of the laws available in English, as well as the definitions or relevant text they contain.

Definitions of genetic testing applied to different legal fields were compared, with the aim to describe its main characteristics (Table 2). Since legal fields often overlap in a single document, more than one legal field may have been identified for each one (Table 3). This multiple marking may also have helped reducing biases in the analysis.

<table>
<thead>
<tr>
<th>LEGAL FIELD</th>
<th>Characteristics</th>
<th>Purpose of GT</th>
<th>Explicitly not (purpose)</th>
<th>Target group of the GT</th>
<th>Example</th>
</tr>
</thead>
<tbody>
<tr>
<td>Labour/insurance law</td>
<td>Identify specific genetic mutations and susceptibilities, for environmental exposure</td>
<td>Screening, diagnosis, treatment, research, only for health purposes</td>
<td>Diagnosis</td>
<td>Employees and insured persons</td>
<td>USA: Executive Order to Prohibit Discrimination in Federal Employment Based on Genetic Information</td>
</tr>
<tr>
<td>Healthcare law</td>
<td>Identify people, human bodies, remains</td>
<td>Specifying patient rights in this field, disclosure of genetic information without permission</td>
<td>Diagnosis in biobanks</td>
<td>Not specified (population, patients, pregnant women)</td>
<td>Australia: Genetic Testing - Guidelines for Prioritizing Genetic Tests</td>
</tr>
<tr>
<td>Forensic/criminal/civil law</td>
<td>Identify people, human bodies, remains</td>
<td>Specifying patient rights in this field, disclosure of genetic information without permission</td>
<td>Diagnosis in biobanks</td>
<td>Strictly defined in the laws (suspected, convicts)</td>
<td>Slovakia: Act on the application of deoxyribonucleic acid for the identification of persons</td>
</tr>
<tr>
<td>Data protection/confidentiality law</td>
<td>Specify patient rights in this field, disclosure of genetic information without permission</td>
<td>Various</td>
<td>Various</td>
<td>Not specified (anyone whose genetic information can be revealed)</td>
<td>Portugal: Personal genetic information and health information</td>
</tr>
<tr>
<td>Data protection/confidentiality law</td>
<td>Specify patient rights in this field, disclosure of genetic information without permission</td>
<td>Various</td>
<td>Various</td>
<td>Various</td>
<td>CoE: Oviedo Convention</td>
</tr>
<tr>
<td>Data protection/confidentiality law</td>
<td>Specify patient rights in this field, disclosure of genetic information without permission</td>
<td>Various</td>
<td>Various</td>
<td>Various</td>
<td>CoE: Oviedo Convention</td>
</tr>
<tr>
<td>Biobanks</td>
<td>Not specified (population, patients, pregnant women)</td>
<td>Strictly defined in the laws (suspected, convicts)</td>
<td>Various</td>
<td>Various</td>
<td>Various</td>
</tr>
<tr>
<td>Complex laws</td>
<td>General</td>
<td>General</td>
<td>General</td>
<td>General</td>
<td>General</td>
</tr>
</tbody>
</table>
Table 3 shows the European (supranational) and national legal norms that were examined. One legal document (from Canada, “Genetic testing and privacy”) could not be imported into NVivo 8, because of technical problems (software incompatibility), in spite of all our efforts.

Table 3 is organized into three groups: (1) “legal fields” refer to the scope of the legal documents (as some fields overlap, they were grouped together for discussion); (2) “type of testing” refers to the context of application of the genetic test; and (3) “object” refers to the type of “material” (in the broadest sense of providing any source of genetic information) that is used for a genetic test. The plus signs in the columns indicate the legal fields touched by each legal document and whether a given term or phrase was used, either in their narrow or broad sense, in it.

The legal documents were also compared based on their legal effect. The proportion of binding and non-binding, supranational and national documents (EU member states and non-EU countries) regarding genetic testing, obtained from the analysis with NVivo8 are shown in Figure 1. Member states issue more binding than non-binding legal documents (56% vs. 44%), while this trend was the opposite in other countries (40% vs. 60%)

Figure 2 shows the cumulative number of binding and non-binding documents, by type and object of genetic testing. The most frequently occurring word/phrase in the documents investigated was DNA (or synonyms). With the exception of DNA, all definitions of genetic testing occurred much more frequently in non-binding legal documents.

![Table 3. Items covered by the various definitions of genetic testing contained in legislation](image-url)
<table>
<thead>
<tr>
<th>Country</th>
<th>Document</th>
<th>Reference</th>
<th>Action</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finland</td>
<td>Privacy in Working Life</td>
<td></td>
<td></td>
</tr>
<tr>
<td>France</td>
<td>Code Penal</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Germany</td>
<td>Predictive Health Information</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Greece</td>
<td>Opinion on Prenatal</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Ireland</td>
<td>A Guide to Ethical Conduct</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Italy</td>
<td>Ethical guidelines</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Latvia</td>
<td>Human Genome Research Law</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Lithuania</td>
<td>The Human Organ Transplants</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Portugal</td>
<td>Personal genetic information</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Slovakia</td>
<td>Identification of persons</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sweden</td>
<td>Act on Genetic Integrity</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Switzerland</td>
<td>Ordinance on DNA</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Australia</td>
<td>Guidelines for prioritaging</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Canada</td>
<td>CCMG Mol.Gen. Guidelines</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Georgia</td>
<td>on the Rights of Patients</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Iceland</td>
<td>... biological samples in biobanks</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Japan</td>
<td>Guidelines for genetic testing</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Norway</td>
<td>Technology in human medicine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Switzerland</td>
<td>The medical use of biotechnology</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

The table above lists various documents from different countries related to genetic information, including laws, guidelines, and reports. Each entry indicates whether the document is relevant (indicated by a check mark) or not.
DISCUSSION

The major general aim of this work was to contribute to the discussion of a consensus definition and of its global applicability. The definitions of genetic testing in legal documents cannot be evaluated without their legal surroundings.

Among the specific aims of WP3.4, was the collection and comparison of definitions of genetic testing contained in legal documents from official bodies in Europe and its Member-States (MS) and that from other countries (USA, Canada, Japan, Australia and others).

Another aim was the comparison of these legal documents regarding genetic testing, based upon those definitions, and the context in which they were used, and to relate them to the specific objectives and purposes of that legislation.

Legal fields and scope of definitions

The legal field is a rank in the vertical structure of the legal system. Each legal field relates to laws or legal norms that are more or less connected, because of their similar topic, issues regulated or legal needs. The legal fields may help to reach more transparent or effective regulation.

The distinction made between the various fields identified in the legal documents was necessarily arbitrary, and is often problematic (Table 2). Furthermore, the documents regulating genetic testing very frequently touch several legal fields at the same time. Also, while, in theory, a distinction can be made between insurance/labour law and privacy/confidentiality, data protection laws, based upon their target groups, in reality these fields are strongly connected and it is difficult to discuss them apart.

For example, the Danish Act No. 286, of 24 April 1996, relates health care data with non-discrimination in employment, and explicitly says that “The purpose of the Act is to ensure that health data are not used wrongfully to limit the possibilities of employees for obtaining or maintaining employment. This shall apply irrespective of whether the data relate to genetic tests, ordinary examinations or come from any other sources.” The same happens with the Finnish Act on the Protection of Privacy in Working Life, which says, on section 15, that “The employer is not permitted to require the employee to take part in genetic testing during recruitment or during the employment relationship, and has no right to know whether or not the employee has ever taken part in such testing”.

Link between legal fields and applications of genetic testing

When definitions of genetic testing were compared, three major broad applications could be identified in these legal documents, for purposes of (1) identification (including forensic, criminal and family law), (2) health care and medical research (health care law), and (3) data protection and antidiscrimination (including insurance, employment, data protection, and privacy and confidentiality law).
1. **Identification**: The definitions contained in the legal documents addressing mainly these applications (identity testing) are often more restrictive. Although forensic genetic testing is also based on the direct investigation of DNA, it usually targets non-coding sequences for (civil or criminal) identification of a single person, rather than testing a specific gene or mutation, as in medical genetic tests.

2. **Medical applications**: These provide often complex definitions, using sometimes coded professional language, and distinguishing among several sub-types of genetic tests. These include tests used for screening, prevention, diagnosis or treatment of a particular hereditary disease or condition, or one with a genetic component.

3. **Data protection and antidiscrimination**: These definitions are usually short and general, lacking categories. In the context of labour or insurance law, definitions of genetic testing aim at well defined groups, while in the case of privacy and confidentiality law they are of much broader application. They usually emphasise that genetic tests should serve only medical or legal purposes, and cannot be used to violate basic human rights or human dignity.

It is important to note that several EU member states have failed, so far, to formulate legislation or other legal norms relating specifically to genetic testing (see Table 1).

Also, very often, there are no general definitions contained or the legal document fails to provide a specific definition of genetic testing. One such example is the Hungarian Health Care Act (1997:CLIV.tv), which does not provide a definition or clarify what it means by a genetic test, although it regulates how genetic testing and counselling should be performed in health care.

![Figure 1. Numbers and proportions of legal documents regarding genetic testing in different types of documents.](image-url)
Comparison of genetic testing definitions in EU and national legal documents

Most legal documents regarding definitions of genetic testing emphasized, at the same time, issues related to health care and privacy; a relatively small number of documents was found within the scope of identification or of insurance/labour law (Table 3). A possible reason for this mixed classification is the importance of privacy of genetic information for several other legal fields.

We could not find any binding documents issued by the EU institutions or the Council of Europe containing definitions of genetic testing (Fig. 1). This is not surprising, since these legal entities have no or only restricted rights to issue binding laws for the Member States. Interestingly, the Member States themselves seem to issue more binding than non-binding legal documents containing genetic testing definitions, while the trend in the non-EU Member States is the opposite: these countries have more definitions of genetic testing in non-binding legal documents. It is important to note, that this difference may be due to the small number of documents available and some major differences in legal systems (countries having a common law system, as the UK and USA, or a continental law tradition, as many European countries).

Examining the types of genetic testing definitions (Fig. 2), we found that the most frequently occurring term/phrase (alone or part of definitions) was DNA (or its synonyms). As a rule, all genetic testing definitions (with the exception of DNA) occurred much more frequently in non-binding legal documents. Considering that the difference in proportion of binding and non-binding documents is small, one may suggest that legislators are aware or at least feel the lack of “consensus definitions”, and this may prevent them from using definitions of genetic testing in binding documents.
Structure of genetic testing definitions: genetic and specific approach

There was no standard for the definitions of genetic testing in the analysed legal documents. Different approaches of the genetic definitions can be identified: some use a general definition, while others apply a more specific one.

Some “general definitions” of genetic testing did not provide a precise description of the purpose and target group at the same time, but a broad approach was applied, often unclear from a medical or scientific point of view. Two types of general definitions of genetic testing could be distinguished among the legal documents examined:

1. The definition includes a scientific description based on medical technical terms, as the role of DNA, genes, etc. As an example, the Australian “Ethical guidelines on the use of assisted reproductive technology in clinical practice and research (as revised in 2007 to take into account the changes in legislation)” states: “A genetic test is one that reveals genetic information. It may be performed on DNA, RNA or protein (the ‘gene product’), or involve measurement of a substance that indirectly reflects gene function.” Another example is Japan’s “Guidelines for Genetic Testing”, which state that: “Genetic testing is aimed at making the diagnosis of a genetic disease and involves the analysis or assays of human DNA, RNA, chromosome, protein (peptide), or metabolite.”

2. Some general genetic definitions list different specific types of genetic testing with the respective technical terms. For example, the EC “Opinion on the ethical aspects of genetic testing in the workplace - Opinion № 18” says: ‘genetic testing’ in this context means the use of a scientific test to obtain information on some aspects of the genetic status of a person, indicative of a present or future medical problem. In the context of employment, “genetic testing” may also incorporate “genetic screening”. An example is the Hungarian “Proposal to the Government on the draft bill on the protection of human genetic data and the rules for genetic tests and research”. It says that a genetic test is “a laboratory test aimed at disclosing DNA and/or chromosome variations and their specific protein products, which are accompanied by or predict effects that have an adverse influence of human health. Types of genetic test include diagnostic, presymptomatic, predictive, heterozygote and prenatal tests. Genetic screening is a wide-range programmed genetic test provided to a population or a group of population for the purpose of identifying certain genetic characteristics in asymptomatic persons, thereafter called collectively as “genetic testing”).

The other main approach is giving specific definitions of genetic testing, highlighting their purpose and target group. The British document on “The Use of Personal Data in Employer/Employee Relationships” declares that “employers should not require employees to undergo genetic testing (or other tests identifying susceptibility to disease) unless it can be objectively justified on either strong public, or employee, health and safety grounds. Such tests may only be carried out with the prior consent of the employee concerned and if the results are interpreted by a qualified health professional who has completed higher specialist training in clinical genetics under the Royal College of Physicians, or an equivalent overseas body”.

It has to be noted that definitions of each type of genetic testing often overlap, since there is no consensus on them. For example, terms as “genetic screening” may be used in a different sense, e.g., population genetic screening or screening for mutations for a given condition or disease in a single
person. Also, presymptomatic and predictive testing may be used either as synonyms, or as different types of genetic tests with different predictive value (to distinguish between tests for high-penetrance monogenic disorders and susceptibility testing for complex diseases). The incoherence among the various definitions is mentioned, for example, in the CoE “Recommendation No. R (92) 3 on Genetic Testing and Screening for Health Care Purposes”, which states that an “essential distinction between genetic diagnosis and genetic screening” must be made.

The Oviedo Convention

The Oviedo Convention has had a crucial role in harmonizing laws on human rights. The “Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine (CETS No. 164)”, known as the Oviedo Convention, was ratified, so far, by 21 European countries. These are: Bosnia-Herzegovina, Bulgaria, Croatia, Cyprus, Czech Republic, Denmark, Estonia, Georgia, Greece, Hungary, Iceland, Lithuania, Moldova, Norway, Portugal, Romania, San Marino, Slovakia, Slovenia, Spain and Turkey.

Article 12 declares that “tests that are predictive of genetic diseases, or which serve 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 counselling”. Thus, all countries that have ratified the Convention have at least one norm regarding presymptomatic, susceptibility and carrier genetic testing, and its need for genetic counselling.

The Additional Protocol concerning Genetic Testing for Health Purposes

But, another important step regarding harmonization of legislation of genetic testing was done very recently, when the Committee of Ministers of the Council of Europe adopted the Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes, in April of 2008.

The Council of Europe focused on the ethical and legal aspects of applications of genetics to health care, especially genetic testing, and emphasised the protection of fundamental human rights. This Additional Protocol also sets down principles relating to the quality of genetic services, prior information and consent, genetic counselling and population screening.

The Explanatory Report of the Additional Protocol (Article 2) declares that it applies to tests carried out for health purposes, which involve analysis of biological samples of human origin, and specifically aim to identify genetic characteristics of a person that are inherited or acquired during early prenatal development.

This Explanatory Report (Article 2) also declares that the notion of “genetic test” is based here on two elements: the method used and the purpose of the test. It is to be understood as a procedure including removal of biological material of human origin, where relevant, as well as the analysis of the personal information obtained there from.
Limitations of the study

There were some limitations with respect to the analysis and sources that may have affected the accuracy of the results.

Language problem. A significant proportion of national legislation and norms was not available in English; therefore, we had to exclude those from the study, since we lacked the resources to translate them.

Availability problem. A general legal database, which includes all the legislation of all European countries and European Institutions, still does not exist at this moment. Most of the legal databases relating to genetics focus on the legislation of the Member States of the European Union.

Differences in legal systems. Differences between continental and common law complicate comparison of the legal documents.

Conclusions

The purpose of this paper was the collection and analysis of legislation on genetic testing and the comparison of the definitions of genetic testing contained in it. Regulation of the process of genetic testing is, however, very diverse and, therefore, this review can only give a general picture of it.

Main legal fields where the genetic testing definitions were identified, included insurance, labour law, privacy, confidentiality, data protection, laws on biobanks, health care and identification (forensic, criminal, family); some laws, often supranational ones, are very complex and deal with various legal fields at the same time.

There was a relationship between the scope of the definitions provided and the legal field to which it pertained, although sometimes this was not very clear.

Most legal documents containing definitions of genetic testing dealt with health care and privacy law at the same time; only a relatively small number of documents from identification or insurance/labour law contained any definitions.

The proportion of non-binding legal documents including a definition of genetic testing was higher than the binding ones (either from EU Member States, or other countries).

The analysis of definitions showed that some are based on a simple scientific explanation and/or list of subtypes of genetic testing. Others are more detailed and refer the target group and/or purpose of testing.

Proper definitions of genetic testing are often missing in national and supranational legal documents dealing with genetic testing and genetic information, in the various legal fields identified.

When present, definitions are extremely variable and, very often, unclear and confusing. Resolving this situation would require a big effort from legislators. Thus, harmonization of these definitions will be very challenging, though also very much needed.
REFERENCES


1. LEGAL DOCUMENTS FROM THE CoE and EU INSTITUTIONS

EC - „25 Recommendations“

**Title:** 25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing


"A broad definition was used for genetic testing, i.e. “any test that yields genetic data”. Genetic data or information relate to inherited or acquired properties that are transmitted during cell division and that affect subsequent generations of offspring (“germinal genetic data”) or cells and tissues (“somatic genetic data”). The Group focused mainly on genetic data transmissible at the germinal level, pertaining to heritable diseases or traits, and not on somatic genetic data which are subject to increasing interest as tools for identification of disease mechanisms and pathways, disease classification and identification of targets for new medicines."

EC - „STRATA Group on Genetic Testing“

**Title:** Ethical, Legal and Social Aspects of Genetic Testing: research, development and clinical applications, STRATA Group on Genetic Testing


"1.2.2 Diagnostic screening

The following types of screening are included in this category:

a) Prenatal screening for major chromosome and neural tube defects is routinely done in most countries using serum tests combined with ultrasound imaging. Confirmation of the diagnosis is carried out using cytogenetic methods involving chromosome analysis.

b) Neonatal screening for treatable diseases such as phenylketonuria (PKU) and hypothyroidism available in all EU countries, mainly through use of low-cost biochemical tests on blood spots taken from new-born babies (known as Guthrie card tests). For other diseases, like galactosemia, screening procedures vary. Neonatal screening for cystic fibrosis has recently been implemented in some countries. In these cases, molecular testing is used to confirm the biochemical test.

c) Carrier screening for cystic fibrosis is recommended and performed in the US, but is not yet being implemented in most European countries, except in cases of male infertility. It has been introduced in the UK and is under consideration in a number of other countries, including France and the Czech Republic."
1.2.3 Predictive testing

The tests in this category cover a broad range of diseases and acquired conditions and can lead to the prediction of the future health status of an individual. However, the dividing line between strictly inherited and acquired conditions is not always clearly defined. Two different groups of predictive tests can be distinguished, based on the nature of the information resulting from them:

a) Presymptomatic tests - The presence of defects in certain specific genes or gene products creates an almost 100% risk of developing a particular disease later in life. Huntington's chorea, Hereditary Polyposis Coli carcinoma (a form of hereditary colon cancer), some rare forms of Alzheimer's disease, and some forms of familial hereditary thyroid carcinoma are the diseases most frequently tested for in genetic centres under this category, representing about 5% of their present workload.

b) Predisposition tests

i) This category includes tests for other disorders in which defects in a single major gene are considered to increase substantially lifetime risk of developing the disease, such as hereditary breast/ovarian carcinoma and HNPCC (hereditary non-polyposis colon carcinoma). In addition, it includes tests for the combined presence of a series of inherited variants of genes or gene products, which increase the risk of developing a particular disease, moderately or significantly – including common multifactorial diseases and traits such as psychiatric disease, cardiovascular disease, diabetes, rheumatoid arthritis, and osteoporosis. With the exception of some tests for genes involved in hemochromatosis, blood clotting or cardiovascular diseases (e.g. Factor V Leiden, and Angiotensin Converting Enzyme), which still require further development, very few clinically useful tests are currently available for this category of disease although some commercial diagnostic kits are on the market. As our knowledge about these diseases increases, this group of multifactorial diseases could potentially become by far the largest target for genetic testing in the future.

ii) Pharmacogenetic tests, which determine the predisposition of individuals to react differentially to drugs, can also be placed in the group of predisposition tests. For some medicines, pharmacogenetic research will ultimately result in doctors being able to select the medicine that is most likely to benefit a particular patient and/or the dose that is most appropriate. At present, pharmacogenetics is not a widespread clinical practice. However, many pharmaceutical companies today routinely include genetic and genomic analyses in clinical medicine development. The topic of pharmacogenetics is covered in full in chapter 5.

iii) Tests for monitoring disease outcome and possible complications are also finding an important application in the diagnosis and/or prediction of the evolution of early disease stages (e.g. in particular cancers). A growing number of applications are slowly moving into clinical practice.

EC - „Opinion of the group of advisers„

Title: Ethical Aspects of Prenatal Diagnosis - Opinion of the Group of Advisers on the Ethical Implications of Biotechnology to the European Commission

URL: http://ec.europa.eu/european_group_ethics/docs/opinion6_en.pdf

"Preconceptional testing or screening allows individuals to be aware of specific genetic risks to their offspring. It could involve the development of simple-to-use genetic testing kits, which raise ethical issues regarding the management of information by the individuals. Preimplantation diagnosis (PID) consists of analysing the genetic components of very early embryos. This allows the transfer of embryos of a particular sex or genotype to the woman’s uterus, thereby greatly reducing the need to consider termination of pregnancy..."
for couples at high risk of transmitting genetic diseases. PID is currently at the experimental stage of clinical trials. Compared with PND, PID, which requires fertilization raises additional ethical questions which the Group intends to study in a later stage.

EC - „Opinion N° 18”

Title: Opinion on the ethical aspects of genetic testing in the workplace - Opinion N° 18

URL: http://ec.europa.eu/european_group_ethics/publications/docs/avis18_compl_en.pdf

“(e) “genetic testing” in this context means the use of a scientific test to obtain information on some aspects of the genetic status of a person, indicative of a present or future medical problem. In the context of employment, "genetic testing" incorporates "genetic screening" and "genetic monitoring";

(f) “genetic screening” in this context means the use of a scientific test to determine whether a person possesses particular variant forms of one or more genes in his/her genome;

(g) “genetic monitoring” in this context means the examination, at regular intervals, for chromosomal abnormalities in samples of cells from a person who may be at risk, in their employment, of exposure to agents which cause genetic damage;”

EC - „Towards quality assurance …”

Title: Towards quality assurance and harmonisation of genetic testing services in the EU

URL: http://www.orpha.net/docs/geneticstesting.pdf

"2.1.1 Working definition

Genetic testing is used to identify variations in the DNA sequence that correlate with a disease or higher risk to develop a disease. This type of test can be used for diagnosis before any symptoms of disease are recognisable and to determine the personal risk for certain multifactorial diseases.

2.1.2 Types of tests

Genetic tests as defined above can be applied serving the following purposes:

• Diagnostic testing: This is the most common reason for a request for a genetic test triggered by a patient presenting clinical signs or symptoms suspected to have a genetic cause. In this case, the test is performed to confirm, refine or exclude a clinical diagnosis. In many cases the test is widely used as an exclusion test with a low probability of a positive diagnosis (an example is a test for fragile-X disease on children with learning difficulty) [3].

• Predictive testing: To estimate the risk to a person with no symptoms of developing a genetic disorder in the future. Usually two forms of predictive testing are distinguished, presymptomatic and predisposition testing:- Presymptomatic testing looks for a mutation (or alteration) in a healthy individual, which, if present, will almost certainly lead to occurrence of symptoms. This type of testing is most applicable to adult-onset genetic conditions like Huntington’s chorea. Adults may have no symptoms of disease at the time of the test but there might be a suspicion of high risk of inheriting a genetic condition from a parent. A minority of individuals in this situation seek predictive information through a genetic test.- Predisposition testing looks for gene mutations
that provide a probability of occurrence of the disorder (e.g. mutations in the genes BRCA 1 and 2 provide
certain susceptibility for breast cancer, but a positive test result does not indicate a 100% risk of developing
appear to be multifactorial, i.e. are susceptible to a number of genetic and environmental influences. The
search for the genetic components of these conditions is currently a major research undertaking, with
enormous commercial implications.

• Carrier testing: Clarification of presence of a gene mutation for a recessively inherited or X-linked disorder
that will not affect the person but could eventually affect his/her relatives. The test result might be important

202. Genetic testing into context for reproductive decisions. Carrier testing in children where the test has no
implication for their own health has been controversial with many geneticists arguing that the possibility of
testing should be delayed until an age when a child can give informed consent for the test.

• Prenatal testing: Clarification if the foetus carries certain mutations or alterations responsible for hereditary
diseases. Prenatal testing for Down syndrome and related conditions usually results from increased risk either
because of maternal age or following a pregnancy screen by ultrasonography or a biochemical test of the
mother’s blood. Prenatal diagnosis for single gene disorders is comparatively rare. It is mainly requested where
parents are at high risk and have direct experience of a serious genetic condition in their own child or in a close
relative. A close liaison between obstetric and genetic services is desirable since parents may choose to end
pregnancies shown to be at high risk of a genetic condition after a test.

• Genetic screening: Predictive testing, prenatal testing and carrier testing can also be offered systematically at
the population level. Genetic screening may be concerned with the general population or with sub-populations
defined on the basis of their risk. Population screening programs are usually decided on and organised by
health authorities at the national or regional level. The only well-established screening programmes are
newborn screening programmes: Testing of new-born shortly after birth for specific disorders such as
phenylketonuria, galactosemia, congenital hypothyroidism. Biochemical genetic testing of newborn infants is
recommended for some monogenic conditions and is carried out in specialist neonatal screening laboratories.

• Preimplantation genetic diagnosis (PIGD) follows an in vitro fertilisation procedure. Genetic testing is carried
out on one or two cells removed from the early embryo. Embryos shown by the PIGD test not to have a genetic
mutation for the condition examined are then implanted in the uterus to attempt to establish a pregnancy. The
scope and range of testing in the context of this study were circumscribed to diagnosis of health effects but
there are many other common applications of these techniques. Pharmacogenetics, which refers to the
identification of DNA variants (polymorphisms) that are related to the variability in drug response, is amongst
these other applications of genetic testing with a potentially massive expansion. Adverse drug responses
currently account for many hospitalisations and deaths per year. On average in Europe, up to 7% of hospital
patients receiving medication experience severe side effects and it is thought that pharmacogenetic testing
could reduce this incidence and may contribute to the development of individualised prescription of medicines,
or the assurance of the 'right medicine for the right patient' [4]. Another type of DNA-based testing is carried
out for disease sub-typing, for example in oncology laboratories to characterize different types of cancer. The
mutations that are under investigation in these cases are most often so-called somatic changes, which arise in a
specific tissue, causing a clone of cells to proliferate. These techniques are also used in the identity testing for
forensic and criminal law applications, to establish or disprove paternity and to confirm family relationships in
immigration applications. This type of testing is currently widespread and occurs in both the public and private
sectors. Areas of related activity that are economically important and share technologies include genetic
testing in non-human areas to promote animal health and food safety and regulation, for example testing for
permissible levels of genetically modified organisms in food products.9
CoE - „Recommendation No. R (92) 3”

**Title:** Recommendation No. R (92) 3 on Genetic Testing and Screening for Health Care Purposes (Feb. 10, 1992)

**URL:** http://www1.umn.edu/humanrts/instree/coerecr92-3.html

"For the purposes of this Recommendation:

a. the term "genetic tests for health care purposes" refers to tests which serve:

- to diagnose and classify a genetic disease;
- to identify unaffected carriers of a defective gene in order to counsel them about the risk of having affected children;
- to detect a serious genetic disease before the clinical onset of symptoms in order to improve the quality of life by using secondary preventive measures and/or to avoid giving birth to affected offspring;
- to identify persons at risk of contracting a disease where both a defective gene and a certain lifestyle are important as causes of the disease;

b. the term "genetic diagnosis" refers to tests carried out to diagnose a presumed ailment on an individual or several members of a family in the framework of a family study;

c. the term "genetic screening" refers to genetic tests carried out on a population as a whole or a subset of it without previous suspicion that the tested individuals may carry the trait.

Footnote: 21. Genetic testing and screening can be carried out at different levels, such as on chromosomes, genes (DNA), proteins, organs or a given individual, and can be complemented with aspects of the family history.

Footnote: 32. The essential distinction between genetic diagnosis and genetic screening is that the latter is not initiated by the individual who is its subject, but by the provider of the screening service."

CoE - „CDBI-CO-GT4"

**Title:** Working Party On Human Genetics (CDBI-CO-GT4), Working document on the applications of genetics for health purposes

**URL:** http://www.coe.int/T/E/Legal_Affairs/Legal_co-operation/Bioethics/Activities/Human_genetics/INF(2003)3e_genetics_working_doc.asp

"Article 15 - Purposes of predictive genetic tests

Tests which are predictive of genetic diseases or disorders or which serve either to identify a person as a carrier of a gene responsible for a disease or disorder, or to detect a genetic predisposition or susceptibility to a disease or disorder may be performed only for health purposes or for scientific research linked to health purposes.

Section IV - Genetic screening for health purposes

Article 20 - Scope of Section IV
The provisions of this section apply to specific tests offered for health purposes in an authorised programme, to an entire population or section of a population in order to identify asymptomatic persons with an increased risk of developing a genetic disease or disorder or transmitting such a disease or disorder to his or her descendants.

Article 21 - Additional criteria to be met before performing screening

A genetic screening programme for health purposes may only be undertaken if the following additional specific conditions are met:

a. The scientific validity of the programme has been established;

b. The programme is recognized for its relevance to health;

c. Effective preventive or treatment measures can be taken in respect of the disease or disorder which is the subject of the screening;

d. Measures shall be taken to adequately inform the population or section of population concerned on the existence, purposes and means of accessing the screening programme;

e. No payment shall be given for participation in the screening programme."

CoE - "OVIEDO Convention"

**Title:** Convention for the Protection of Human Rights and Dignity of the Human Being


"Article 11 – Non-discrimination

Any form of discrimination against a person on grounds of his or her genetic heritage is prohibited.

Article 12 – Predictive genetic tests

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."

**Title:** Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes

**URL:** [http://www.bioeticaweb.com/content/view/4603/858/lang.es/](http://www.bioeticaweb.com/content/view/4603/858/lang.es/)

Article 2- Scope

This Protocol applies to 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 which are inherited or acquired during early prenatal development (hereinafter referred to as “genetic tests”).
2. LEGISLATION FROM EU MEMBER STATES

Belgium - „Royal Decree”

Title: Royal Decree
URL: http://www.coe.int/t/e/legal_affairs/legal_co-operation/bioethics/news/Human%20Genetics1.asp

"Defined by a Royal Decree in 1987 (article 1) as: “Centers where diagnoses are made on”:
1. the possibility that malformation or abnormalities, either mental or physical, are determined by heredity
2. the nature of in (1) mentioned malformation or abnormalities
3. the carrier state of hereditary properties

In the explanatory memory of this Decree is stated that “Activities of centers of hereditary diseases are often so complex, the investigated anomalies so rare making a limitation of numbers of such centers mandatory”.

Bulgaria - „Bulgarian Health Act”

Title: Bulgarian Health Act
URL: http://www.who.int/idhl-rils/idhl/Bulg06001a.pdf

"activities, directed to: 1. prophylactic and diagnostic investigations for proving and classification of genetic diseases; 2. dispensary system for the persons with increased risk of occurrence and development of genetic diseases; 3. healing of inherited diseases, innate anomalies and predisposition; 4. establishing of inherited characteristics and identification of parent; 5. preservation of genetic information. Art. 138. Prophylactic genetic investigations shall be implemented for: 1. determining the risk of occurrence of genetic disease in the generation; 2. identification of clinically healthy carriers of genetic deviations; 3. diagnostics of inherited and other diseases in the periods before and during the pregnancy and after the childbirth."

Cyprus - „The Safeguarding …”

Title: The Safeguarding and Protection of Patients’ rights Law, 2004
URL: http://www.who.int/idhl-rils/idhl/Cypr.05002.pdf

Genetic data means all data, whatever type, concerning the hereditary characteristics of an individual or concerning the pattern of inheritance of such characteristics within related groups of individuals

Denmark - „... on patients’ rights”

Title: Law on Patients Rights, 1999, 50 (1) - Health Law no. 482, of 1 July 1998, on Patients’ Rights
URL: http://waml.haifa.ac.il/index/reference/legislation/denmark/denmark1.htm

"DENMARK. law No. 482 of 1 July 1998 on patients’ rights 5. (1) No medical check connected with the closure or amendment of an insurance contract shall involve questioning the proposer’s blood relatives or, unless the
condition is manifest the proposer, regarding a medical condition, as referred to in clause a of subsection 2 of Section 3, insofar as the condition is hereditary, or regarding tests which the proposer or the proposer’s blood relatives have undergone with a view to determining genetic predisposition to a medical condition or conditions, or regarding the results of such tests, unless the sum insured exceeds the enquiry limit. In the assessment of a proposal regarding the closure or amendment of an insurance contract, and in the course of any related medical check, no use may be made of genetic information held by the commissioning party, the performing physician or the medical adviser, regarding the proposer or the proposer’s blood relatives, which has not been obtained in connection with that proposal."

**Denmark - „Law on artificial fertilization…”**

*Title: Law on artificial fertilization in connection with medical treatment, diagnosis, and research (Research on embryonic stem cells)*


"25. (1) Biomedical experimentation on fertilized human oocytes and gametes intended for use in fertilization may only be carried out in the following cases: 1. for the purpose of improving in vitro fertilization or similar techniques intended to bring about pregnancy; 2. in order to improve techniques for the genetic testing of a fertilized oocyte with a view to establishing the possible presence of a serious hereditary disease or an important chromosome abnormality (pre-implantation diagnosis); and 3. if the purpose of experiments involving the use of fertilized oocytes and stem cells derived therefrom is to obtain new knowledge that could improve the possibilities of treating diseases in human beings. (2) The removal and fertilization of an oocyte in order to carry out experiments other than those referred to in subsection 1 shall be prohibited."

**Denmark - „Act on the use of health data…”**

*Title: Act on the use of health data etc. on the labour market*

*URL: [http://www.bm.dk/graphics/Dokumenter/English/helbredsoplysningslov.pdf](http://www.bm.dk/graphics/Dokumenter/English/helbredsoplysningslov.pdf)*

Act No 286 of 24 April 1996. 1. - (1) The purpose of the Act is to to ensure that health data are not used wrongfully to limit the possibilities of employees for obtaining or maintaining employment. This shall apply irrespective of whether the data relate to genetic tests, ordinary examinations or come from any other sources.

**Estonia - „Human Genes Research Act”**

*Title: Human Genes Research Act*


"§ 6. Genetic research and genetic testing (1) Genetic research relating to the Gene Bank is permitted in order to study and describe the links between genes, the physical and social environment and the lifestyles of people, to find medicinal products or methods of treatment on the basis thereof, to assess individual health hazards and to prevent illnesses. (2) Genetic testing to which the provisions of Chapters 2, 3 and 4 of this Act do not apply may be performed pursuant to the procedure and for the purposes provided by law. Tissue samples
taken from people in the course of genetic testing and the results of research performed on such samples shall not be added to the Gene Bank."

**Finland - „Privacy in Working Life”**

**Title:** Act on the Protection of Privacy in Working Life


Section 15 - Genetic testing The employer is not permitted to require the employee to take part in genetic testing during recruitment or during the employment relationship, and has no right to know whether or not the employee has ever taken part in such testing.

**France - „Penal Code”**

**Title:** Penal Code

**URL:** http://www.legifrance.gouv.fr/html/codes_traduits/code_penal_textA.htm#SECTION%206.%20-%20OFFENCES%20AGAINST%20PERSONS%20RESULTING%20FROM%20EXAMINATION%20OF%20GENETIC%20CHARACTERISTICS%20OR%20IDENTIFICATION%20OF%20GENETIC%20IMPRINTS

The diversion from its medical or scientific research objectives of information collected on a person by way of a study of his genetic characteristics is punished by one year's imprisonment and a fine of € 15,000.

**France - „Criminal Procedure”**

**Title:** Criminal Procedure

**URL:**

"Article 706-54, (Law n° 98-648 of 17 June 1998 art. 28 Official Journal of 18 June 1998)(Law n° 2001-1062 of 15 November 2001 art. 56 Official Journal of 16 November 2001) A national automated data bank is created to centralize the genetic traces as well as the genetic fingerprints of the persons convicted of any of the offences set out in article 706-55, so as to facilitate the identification of and search for the perpetrators of these offences. This data bank is placed under the supervision of a judge. The terms of implementation of the present article, including the time during which the recorded information may be preserved, are determined by Decree of the Council of State, taken after hearing the opinion of the National Commission for Computer Science and Civil Liberties. The genetic fingerprints of the persons against whom there is serious and corroborating evidence liable to justify their placement under judicial examination for one of the offences covered in article 706-47, may be compared with the data recorded in the data bank at the request of the investigating judge or district prosecutor. They may not, however, be recorded in this data bank."
"For the purposes of this Opinion, genetic tests are defined as all tests that directly furnish information on an individual's genetic endowment by means of the analysis of substances taken from the body. These may be tests of chromosomes (cytogenetic analysis), DNA or RNA (molecular genetic analysis) or gene products (biochemical or immunochemical assay). Depending on the presence or absence of pathological manifestations, the procedure will constitute either genetic diagnosis or a predictive genetic test. Phenotypic examinations (e.g. analysis of an individual's externally visible characters) or imaging techniques, on the other hand, are not regarded as genetic tests, even if information on genetic characters can be derived from them in certain cases. A predictive genetic diagnosis may be appropriate where someone is at increased risk of developing a genetically mediated disease owing to its occurrence in his family and to the laws of inheritance. If the disease has a simple hereditary pathway and genetic diagnosis shows that the subject is not a carrier of the pathological genetic alteration, the possibility of his being affected by this hereditary disorder can be definitively ruled out. If the genetic diagnosis reveals a pathological mutation, the probability of illness depends on the mutation's penetrance. Hence a direct genetic diagnosis, as opposed to the familial history, has the consequence that the subject is assigned to a different risk group, because he now carries not the familial average risk but the individually ascertained risk. Depending on the disorder concerned, the extremes may be 0% or 100%.

Today, numerous diseases and pathological predispositions can be attributed (at least in part) to a chromosome change or variation affecting single genes and their regulation. Hence, cytogenetic and molecular genetic analyses are becoming increasingly important. Compared with traditional diagnostic tools, they often facilitate the diagnostic process because the test can usually be performed using a blood sample from the patient. In addition, these analyses sometimes also provide a more precise diagnosis (e.g. compared with family tree analyses), allow a more detailed demonstration of heterozygosity and help detect pathological predispositions. Cytogenetic and molecular genetic testing procedures are applied in medicine in different contexts and for different purposes, both within the field of human genetics and outside (see Chart 6 below). These include in particular:

— confirmation of the diagnosis in clinically manifest hereditary disease (diagnostic genetic testing),

— diagnosis of a predisposition for a given disease before its outbreak (predictive genetic testing),

— prenatal diagnosis of conspicuous chromosomal features and specific single gene changes allowing conclusions to be drawn concerning possible disabilities or diseases of the expected child (prenatal genetic analysis),

— diagnosis of specific conspicuous chromosomal and molecular genetic features in embryos produced in vitro before their transfer to the uterus (preimplantation genetic analysis),
— examination of female germ cells before their fertilisation (preconceptional diagnosis),
— screening not just of individuals but of the entire population or of sections of the latter (genetic screening),
— assessment of genetically induced differences in the response of patients to active drugs (pharmacogenetic diagnosis).

Germany - „Genetic diagnosis …”

Title: Genetic diagnosis before and during pregnancy
URL: http://www.ethikrat.org/_english/publications/Stn_PID_engl.pdf

"Pre-implantation genetic diagnosis is defined as the genetic examination of embryos produced by extracorporeal fertilization when they are a few days old. Out of a number of embryos, the ones selected for transfer to the woman’s uterus are those for which certain chromosomal disorders and/or mutations can be ruled out with a high degree of probability. A number of precautionary examinations are carried out during pregnancy. The guidelines adopted by the Bundesausschusserärzte und Krankenkassen [Federal Committee of Physicians and Health Insurance Funds] (the “Maternity Guidelines” 5) are intended to ensure that treatment is provided in accordance with the rules of the art of medicine and having regard to the generally acknowledged state of medical knowledge. The precautionary examinations are conducted not only on the expectant mother herself – involving, for example, blood tests and gynaecological examinations – but also on the unborn child."

Greece - „Opinion on Prenatal …”

Title: Opinion on Prenatal and Pre-Implantation Diagnosis
URL: http://www.bioethics.gr/media/pdf/recommendations/1_pd_pgd_opin_eng2.pdf

1. Prenatal Diagnosis “Prenatal diagnosis” (PD) signifies the testing of the embryo in vivo with certain methods in order to identify in time potential abnormalities or diseases. These methods are non-invasive (e.g. ultrasonography, maternal blood tests) or invasive (amniocentesis, trophoblast testing). In the case of invasive methods embryo cells are harvested by puncture from the amniotic fluid or from the trophoblast. Invasive prenatal tests detect severe chromosomal abnormalities (e.g. Down Syndrome) and genetic conditions (e.g. thalassemia, cystic fibrosis) and also non-pathological phenotypic characteristics (e.g. the embryo’s sex). They are usually conducted during the first trimester of pregnancy. 2. Pre-implantation diagnosis “Pre-implantation genetic diagnosis” (PGD) means testing in vitro embryos that were created in the context of assisted conception. This test can identify severe abnormalities and diseases as well as the embryo’s gender. It is performed on 1-2 blastomeres that are removed from the embryo without destroying it. Pre-implantation diagnosis is not yet applied universally in assisted conception, as it requires specialized laboratory equipment and a matching level of expertise.

Greece - „Use of Genetic Data”

Title: Recommendation on the Collection and Use of Genetic Data
Respect for the value of human beings requires the free and informed consent of the person whose biological sample is collected for the purpose of genetic testing.

**Hungary - „Health Care Act”**

**Title:** Health Care Act


Definition: 3. § ka) screening: this type of exploration aims to identify a contingent illness of symptom-free persons in the early phase of diseases or the premorbus status including those hazardous effects which make susceptible to morbid.

**Hungary - „Proposal to the Government...”**

**Title:** Proposal to the Government on the draft bill on the protection of human genetic data and the rules for genetic tests and research

**URL:** [http://www.ett.hu/tajekoztato/humgen.pdf](http://www.ett.hu/tajekoztato/humgen.pdf)

"h) genetic test: a laboratory test aimed at disclosing DNA and/or chromosome variations and their specific protein products, which are accompanied by or predict effects that have an adverse influence of human health. Types of genetic test: diagnostic, Presymptomatic, predictive tests, heterozygote tests, prenatal tests. i) genetic screening: a wide-range programmed genetic test provided to a population or a group of population for the purpose of identifying certain genetic characteristics in asymptomatic persons (paragraphs h-i) hereinafter collectively: genetic testing); "

**Ireland - „A Guide to Ethical Conduct ...”**

**Title:** A Guide to Ethical Conduct and Behaviour (6th edition)


Genetic testing may be of benefit in diagnosing an illness or predicting its development in the future. Individuals who undergo such testing should be counseled regarding the consequences of their actions and testing should not be done without their informed consent.

**Ireland - „Disability Act 2005”**

**Title:** Disability Act 2005

“genetic testing” means the examination of samples taken from a living person for the purpose of analyzing the person’s deoxyribonucleic or ribonucleic acid by means of chromosomal analysis or by any other means for the purpose of—(a) confirming the identity or nature of an existing symptomatic disease, (b) ascertaining whether the person has a genetic predisposition or susceptibility to a disease, or (c) identifying the carrier of a disease; “processing” has the meaning assigned to it by the Acts.

**Italy - “Bioethical guidelines ...”**

*Title:* Bioethical Guidelines for Genetic Testing

*URL:* [http://www.palazzochigi.it/bioetica/eng/opinions/geneticstest.html](http://www.palazzochigi.it/bioetica/eng/opinions/geneticstest.html)

“Genetic tests are commonly defined as “the analysis of specific genes, their product or their function, as well as of any other type of study of the DNA or chromosomes, aimed at identifying or ruling out DNA modifications presumably linked to genetic pathologies”.”

**Latvia - „Human Genome Research Law”**

*Title:* Human Genome Research Law


“12) genetic testing – gene analysis of a person performed for the purpose of the identification of the person or the diagnosis of diseases, as well as for the selection of prophylactic or treatment measures;”

**Northern Ireland - „The Human Organ Transplants”**

*Title:* The Human Organ Transplants (Establishment of Relationship) Regulations Statutory Rule 1998 No. 389


““genetic tests” means tests based on DNA variations”

**Portugal - “Personal genetic information”**

*Title:* Personal genetic information and health information


"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 heterozygous 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.

Slovakia - „...identification of persons”

Title: Act on the application of deoxyribonucleic acid for the identification of persons

URL: http://www.minv.sk/en/_private/417.htm

"§ 2 Definitions

For the purpose of this Act the following definitions are introduced

a) the sample is a biological material coming from the human body,

b) the analysis of deoxyribonucleic acid is the process of the sample analysis by the methods of molecular biology and genetics executed from the non-encoding sections of the deoxyribonucleic acid molecule that do not contain information on specific hereditary properties,

c) the deoxyribonucleic acid profile is the result of deoxyribonucleic acid analysis in the form of alpha-numeric code,

d) the ensured sample is the sample ensured on the place of criminal offence or on other place that is related to the criminal offence, the sample ensured in connection with the searching of missing person and in connection with the identification of person,

e) identification of person is the individual identification of person, dead body or single parts of human body.”

Sweden - „Biobanks in Medical Care Act”

Title: Biobanks in Medical Care Act

URL: http://www.sweden.gov.se/content/1/c6/02/31/26/f69e36fd.pdf

not applying for in the course of medical care for analysis
Sweden - “Act on Genetic Integrity”

Genetic information: information about the result of a genetic investigation, excluding that part of information which is connected with factual health status

prenatal diagnosis: medical investigation of a pregnant woman and the fetus she is carrying

Gene therapy: a treatment which means that a healthy copy of a gene with help of a carrier (vector) is introduced in a cell from an individual who has a genetic disease

Gene therapy: a treatment which means that a healthy copy of a gene with help of a carrier (vector) is introduced in a cell from an individual who has a genetic disease

The Netherlands - “The application of Genetics ...”

Title: Policy Document “The application of Genetics in the Health Care Sector”


"DNA diagnosis - Testing for changes and variants in the DNA that are associated with the occurrence of, the risk of and the course of certain syndromes or with the reaction to certain medical treatments.

Genetic screening - Genetic testing without an individual indication, based on its being made available to a predetermined group of the population by the medical profession, the government etc.

Clinical genetic testing - Testing for hereditary and congenital abnormalities. Numerous methods are now available, among them DNA diagnosis, chromosome testing, biochemical testing, echoscope testing and family history research. In principle testing of this kind can be carried out at any stage of life.

Preimplantation genetic diagnosis (PGD) - Testing for genetic abnormalities of an embryo created in vitro. Only embryos in which the genetic abnormality concerned is not found are considered for implantation in the woman’s womb.

Prenatal genetic testing - Testing for genetic abnormalities during pregnancy.

Presymptomatic diagnosis - Diagnosis that is made at a time when the individual being examined is not (yet) ill. The purpose is to discover whether or not an individual is a carrier of a particular genetic abnormality.

Predictive genetic testing - Testing focused on the detection and reduction of risks of genetic disorders later in life. Genetic abnormalities can be determined on the basis of a genetic test, and the chances of these abnormalities actually being expressed can be calculated." §5 genetic investigation: an investigation within the health care system or medical research which is intended to give information about a human being's inheritance by molecular, microbiological, immunological, biochemical, cytogenetic or other equivalent method, or by collecting information about the persons or his/her biological relatives.
"Embryo testing 63. The 1990 Act as it stands does not specifically mention embryo testing and currently confers a wide discretion on the HFEA to make licensing decisions on this issue. The Bill adds paragraphs 1ZA to 1ZC to Schedule 2 of the 1990 Act dealing with embryo testing (for example preimplantation genetic diagnosis of an hereditary disease), and practices designed to secure that a resulting child will be of one sex rather than the other."

"Genetic Test - A test to detect the presence or absence of, or change in, a particular gene or chromosome."

"8.23 A genetic predisposition can take the form of an increased likelihood of developing a health condition in the future. There is some concern that people with a genetic predisposition may be discriminated against by employers and insurers.8.24 Until recently, tests were available only for a very limited number of single gene inherited disorders or ‘familial’ forms of diseases such as cancer or heart disease. However, increased knowledge of the role of genetics in health will inevitably increase the role of genetic tests in many areas of medicine. However, it is possible that genetic testing may be used to assess long term health prospects in pre-employment health checks in the future. This could lead to unfair discrimination against those with a presymptomatic genetic condition. For example, applicants with a high risk of illness in the future might be denied employment because of their chances of early retirement or multiple sickness leave."
"4. Genetic testing is in its infancy and its long term implications for insurance, preventative medicine or treatment is indeterminate. The majority of genetic tests confirm diagnoses of ill health and inform treatments. Such diagnostic testing falls into the same category as other clinical technologies."

United Kingdom - „The use of Personal Data …”

**Title:** Draft Code of Practice: The use of Personal Data in Employer/Employee Relationships

**URL:** [http://www.fedee.com/dpcp.html](http://www.fedee.com/dpcp.html)

"51: Employers should not require employees to undergo genetic testing (or other tests identifying susceptibility to disease) unless it can be objectively justified on either strong public, or employee, health and safety grounds. Such tests may only be carried out with the prior consent of the employee concerned and if the results are interpreted by a qualified health professional who has completed higher specialist training in clinical genetics under the Royal College of Physicians, or an equivalent overseas body."

United Kingdom - „Our Inheritance, Our Future”

**Title:** Our Inheritance, Our Future: Realising the Potential of Genetics in the NHS


""Genetic testing
1.6 At present, most NHS genetics work is concerned with inherited disorders caused by a defect in a single gene or chromosome that results in a specific medical condition or syndrome."

United Kingdom - „..., genetic paternity testing”

**Title:** Code of Practice and Guidance with Respect to Genetic Paternity Testing Services


"Modern paternity testing often involves comparing the DNA of individuals to determine their biological relationship. Tests are undertaken to determine, with a high level of accuracy, whether a man is the biological father of the child in question. The scientific testing methods used to perform DNA based paternity testing are constantly evolving and improving."
"3.1 The term “genetic testing” is used in different ways (see table 1): (a) Testing for inherited disease. Methods include DNA analysis, chromosome analysis, biochemical or immunological tests or even radiology. This covers genetic tests that potentially identify significant risks of inherited or congenital disorders for individuals and their families. Currently these are predominantly the single gene disorders including the inherited cancer predisposition disorders (particularly breast and colon cancer, a small percentage of these common cancers being associated with single gene changes highly predictive of disease development)."

"Purposes of general application5 (1) Use of the results of an analysis of DNA for any of the following purposes is use for an excepted purpose— (a) the medical diagnosis or treatment of the person whose body manufactured the DNA; (b) purposes of functions of a coroner; (c) purposes of functions of a procurator fiscal in connection with the investigation of deaths; (d) the prevention or detection of crime; (e) the conduct of a prosecution; (f) purposes of national security; (g) implementing an order or direction of a court or tribunal, including one outside the United Kingdom."
3. LEGISLATION FROM OTHER COUNTRIES (non-EU)

Australia - „Guidelines for prioritizing...“

Title: Genetic Testing - Guidelines for Prioritizing Genetic Tests


"High Priority

1. Prenatal Testing

- Where the confirmation of a clinical diagnosis by molecular testing will assist parents who may use the information in making reproductive choices.
- Where the confirmation of the clinical diagnosis will enable treatment options to be instituted which might be early in the newborn period.
- Where gonadal mosaicism is recognized to occur frequently (eg Osteogenesis Imperfecta with a risk of 3 – 4%)

2. Diagnostic Testing

- When confirmation of a clinical diagnosis will restore reproductive confidence in the family.
- When confirmation of a clinical diagnosis will lead to changes in management of an affected person.
- Where a diagnostic test can lead to predictive testing of other at-risk family members.
- To confirm a clinical diagnosis where it is relevant to screening for disease complications.
- To confirm a clinical diagnosis where it is relevant for funding purposes eg extra aid at school.

3. Carrier Testing

- Where the patient has had genetic counselling and is aware of a high likelihood of being a carrier based on family history or ethnicity and the patient has accepted the advantages and limitations of carrier testing.
- When there are prenatal diagnosis implications for a family because of a known family history.
- Where one partner is a known carrier of a recessive condition and carrier testing of the other partner may lead to the possibility of prenatal diagnosis and accurate reproductive counseling.

4. Presymptomatic and Predictive Testing

- Where there is a known family history of a disorder and mutation is known.
- Where there is definitive testing available and there is a family history of the disorder ie Huntington disease.

Low Priority

1. Prenatal Testing

- Where confirmation of a clinical diagnosis by molecular testing will not alter the reproductive choices or obstetric or perinatal care for the patient.
- Where there is only a low theoretical risk of gonadal mosaicism.
- Where there is a recessive condition and there is no need for carrier testing for the new partner who is at a low risk of being a carrier.

2. Diagnostic Testing
• Where the clinical diagnosis is confirmed by other means and genetic testing will not alter the patient’s management or options.
• Where the test has been requested by the parents or health professionals and the geneticist thinks a diagnosis is unlikely or the test is not clinically indicated.
• Where confirmation of the clinical diagnosis by genetic testing will not influence whether prenatal testing is undertaken and or the type of test.
• Where the genetic test will not lead to confirmation or predictive testing of other family members eg no at risk relatives.
• Where confirmation of the clinical diagnosis will not alter screening of potential disease complications.

3. Carrier Testing

• Where the disorder is rare and there is no family history.
• Where the testing will not alter the lifestyle or health options for a person.

4. Presymptomatic and Predictive Testing

• Where there is no family history of the disorder.
• Where the only people to have predictive testing would be children for adult onset disorders.”

Australia - „Ethical guidelines...”

Title: Ethical guidelines on the use of assisted reproductive technology in clinical practice and research (as revised in 2007 to take into account the changes in legislation)


“PGD is currently used to detect serious genetic conditions, to improve ART outcomes and, in rare circumstances, to select an embryo with compatible tissue for a sibling.”

Australia - „Genetic testing and storage...”

Title: Ethical Aspects of Human Genetic Testing: an Information Paper


“A genetic test is one that reveals genetic information. It may be performed on DNA, RNA or protein (the ‘gene product’), or involve measurement of a substance that indirectly reflects gene function.

2.1 Types of genetic test

2.1.1 Tests to make a diagnosis in a person who has features of a genetic disorder. Diagnostic test—a test performed to make or confirm a diagnosis of a specific disorder in a person who already has symptoms and/or signs.

2.1.2 Tests to determine the presence or absence of a genetic variant, or variants, in a person who has no features of the disorder at the time of testing, in order to use the information to predict the likelihood that the person will develop the disorder in the future. (a) Presymptomatic test—a test performed on a person who has
no symptoms of a specific disorder at the time of testing, to determine whether or not he/she has a mutant gene. (b) Predictive test—a test performed on a person who has no symptoms of a specific disorder at the time of testing, to determine whether or not he/she has a mutant gene. (c) Susceptibility test—a test performed on a person who has no symptoms of a specific disorder at the time of testing, to determine whether or not he/she has a genetic variant or variants which increase the likelihood (the risk increases are often small) that the person will develop symptoms of the disorder in question at some time in the future.

2.1.3 Tests performed to determine the presence or absence of a genetic variant which will not cause a disorder but which, if transmitted, may be associated with the disorder in offspring. Carrier test—a test performed on a person to determine whether or not he/she has a mutated gene or chromosome abnormality which will not affect the person’s health, but increases his/her chance of having children with the disorder in question.

2.2 Other times when tests are performed

Prenatal test — a test, usually diagnostic or Presymptomatic, carried out on a developing foetus.

Pre-implantation test — a form of Presymptomatic test carried out on early embryos in the laboratory, with a view to transferring to the mother’s uterus only those which will not develop the disorder in question.

Screening test — A screening test is one that is performed on individuals not known to be at increased risk of a particular disorder that is, those with no family history, symptoms or other reason to suggest an increased risk.”

Australia - „ALRC 96”

Title: ALRC 96 Essentially Yours: The Protection of Human Genetic Information in Australia, This Report reflects the law as at 14 March 2003

URL: http://www.austlii.edu.au/au/other/alrc/publications/reports/96/10_Genetic_testing.doc.html#heading1

“Some genetic tests (here called DNA tests) directly analyze DNA or RNA. For example, testing of a genetic sequence may be undertaken by targeting a segment of DNA or RNA using a process known as polymerase chain reaction (PCR). Some tests do not analyze DNA or RNA material directly but test the biological products of particular genes. The measurement of certain proteins produced by genes, or certain metabolites, may reveal valuable information about gene function itself. Some routine biochemical tests of non-genetic substances may also reveal genetic information. Some genetic diagnoses are made on the basis of the morphological characteristics of certain cells, tissues or at postmortem examination. Finally, some medical imaging processes reveal important genetic information.”

Canada - „Genetic testing and privacy”

Title: Genetic testing and privacy

URL: http://www.privcom.gc.ca/information/02_05_11_e.pdf

“The general term “genetic testing” can be divided into categories: genetic screening, genetic monitoring and forensic DNA analysis (sometimes colloquially called genetic or DNA fingerprinting”). “Genetic testing” here will refer to these three types of tests collectively unless the context shows that the particular type of testing in intended. Genetic screening - Genetic screening presents a snapshot of one’s genetic makeup at a given time.
Genes, however, can mutate. Therefore a test taken a long ago may not accurately identify one's genetic makeup. Genetic monitoring - Genetic monitoring is the periodic examination of individuals (such as employees or persons living near chemical dumps or nuclear facilities) to find early indications of genetic mutations. Genetic monitoring can serve two purposes. First, it can identify changes in an individual's genetic makeup that require a remedy. This might include treatment or removal from the environment to prevent further mutations. Second, monitoring of a group could identify environmental hazards (in a paint shop or a chemical factory, for example) that need to be reduced or eliminated. Forensic DNA analysis - Unlike monitoring or screening, forensic DNA analysis does not seek to identify genetic disorders or changes in genetic structure. In short, it is not a diagnostic tool. Instead, it looks for a match or a relationship between two genetic samples. A specific DNA pattern can be associated with a specific individual, much like fingerprints."

Canada - „CCMG Mol.Gen. Guidelines”

Title: CCMG Molecular Genetics Guidelines


"Analysis of DNA and/or RNA to detect heritable disorders as well as germline mutations that confer an increased susceptibility to a disease. This includes prenatal, diagnostic, presymptomatic, carrier and susceptibility testing. These guidelines do not apply to the detection of acquired genetic changes."

Georgia - „on the Rights of Patients”

Title: Law of Georgia of 5 May 2000 on the Rights of Patients

URL: http://www.aitel.hist.no/~walterk/wkeim/files/Georgia_Patients_Rights_Law.htm

“CHAPTER VI - RIGHTS IN THE FILED OF GENETIC COUNSELLING AND GENE THERAPY

Article 31 - Discrimination against a person on grounds of his/her genetic heritage is prohibited.

Article 32 - Tests, which serve to identify a gene responsible for a disease or to detect a genetic predisposition to a disease, shall be carried out only for: a) patient’s healthcare purposes; b) scientific research linked to health purposes.

Article 33 - Any intervention seeking to modify the human genome shall only be carried out for diagnostic, therapeutic or preventive purposes and only if its aim is not to modify the genome of patient’s descendants.

Article 34 - The use of methods of medically assisted procreation for the purpose of sex selection shall be prohibited, with the exception of the cases when hereditary sex-related disease is to be avoided."

Iceland - „... biological samples in biobanks”

Title: Regulations on the keeping and utilization of biological samples in biobanks

URL: http://eng.heilbrigdisraduneyti.is/laws-and-regulations/nr/684
Clinical test: test carried out in order to provide health service to an individual.

Japan - „Guidelines for genetic testing“

Title: Guideline for Genetic Testing

URL: http://jshg.jp/pdf/10academies_e.pdf

"These guidelines concern genetic testing (chromosome analysis, biochemical testing and DNA-based testing) for gene mutations, chromosomal aberrations or their related germline abnormalities. The tests include those for clinical diagnosis, carrier detection, Presymptomatic diagnosis, disease susceptibility estimation (including so-called diathesis diagnosis), pharmacogenetic diagnosis, prenatal diagnosis, and newborn screening for inborn errors of metabolism. However, the guidelines do not cover tests for gene mutation, gene expression and chromosome abnormality which are confined to somatic cells such as cancer cells nor those for infectious agents, e.g., bacteria and viruses, and DNA testing for forensic medicine such as determination of parentage (paternity testing)."

Japan - „using DNA analysis“

Title: Guidelines for Genetic Testing Using DNA Analysis

URL: http://eubios.info/EJ64/EJ64I.htm

In addition to the purpose of confirmation of known and expressed diseases, genetic testing is also performed for purposes of carrier detection, Presymptomatic diagnosis and prenatal diagnosis in the counselee and families, who may be asymptomatic, based on the genetic information obtained. Genetic testing can directly determine the gene mutation responsible for the disease, or can indirectly find the causative genetic elements with high probability using DNA polymorphism. When conducting these diagnoses, appropriate methods must be chosen, depending upon the disease, genetic information, and the collected samples. In genetic testing the human rights of the person who is examined (henceforth called the subject), and his/her family, must be protected, and proper and appropriate genetic testing should be promoted.

Norway - „biotechnology in human medicine“

Title: Act of 5 December 2003 No. 100 Relating to the Application of Biotechnology in Human Medicine, etc

URL: http://www.bion.no/lov/Biotechnology_act_MASTER.pdf

"§ 5-1. Definitions - For the purpose of this Act, genetic testing means all types of analyses of human genetic material at both nucleic acid and chromosome level, analyses of genetic products and their function, and examination of organs to obtain information on human genetic constitution. For the purpose of this Act, postnatal genetic testing means :a) genetic testing to diagnose a disease, b) Presymptomatic genetic testing, predictive genetic testing and testing to determine whether or not a person is a carrier of hereditary disease that will only be expressed in later generations, c) laboratory genetic testing to determine sex, with the exception of laboratory genetic testing for identification purposes."
§ 2-14. Genetic testing of embryos - Genetic testing of embryos before implantation into the womb, including tests designed to choose the sex of the child (pre-implantation genetic diagnosis) may only be carried out in special cases of serious sex-linked hereditary diseases for which no treatment is available."

**Norway - „The medical use of biotechnology“**

*Title:* The medical use of biotechnology  
*URL:* [http://www.who.int/idhl-rils/frame.cfm?language=english](http://www.who.int/idhl-rils/frame.cfm?language=english)

"Chapter 5. Prenatal diagnosis  
5-1. Definition For the purposes of this Law, 'prenatal diagnosis' means: (a) the examination of the fetus or pregnant woman in order to detect or rule out the possibility of genetic disease or developmental abnormalities in the fetus; and (b) ultrasonic examination of the pregnant woman.

5-2. Approval of prenatal diagnosis The types of examination and methods, pursuant to item (a) of Section 5-1, for the purpose of detecting or ruling out the possibility of genetic disease or developmental abnormalities in the fetus, shall be subject to approval from the Ministry."

**Switzerland - „Ordinance on DNA“**

*Title:* Ordinance of 31 May 2000 on the Information System Based on DNA Profiles (Ordinance on DNA)  
*URL:* [http://www.who.int/idhl-rils/frame.cfm?language=english](http://www.who.int/idhl-rils/frame.cfm?language=english)

"Sec. 3 defines "'DNA profile'" as an alphanumeric code established, with the aid of molecular biology techniques, from the non-coding sequences of DNA genetic material"

**United States - „Executive Order ...“**

*Title:* Executive Order to Prohibit Discrimination in Federal Employment Based on Genetic Information  

"Definition of genetic monitoring: Genetic monitoring means the periodic examination of employees to evaluate acquired modifications to their genetic material, such as chromosomal damage or evidence of increased occurrence of mutations, that may have developed in the course of employment due to exposure to toxic substances in the workplace, in order to identify, evaluate, respond to the effects of, or control adverse environmental exposures in the workplace. Genetic test means the analysis of human DNA, RNA, chromosomes, proteins, or certain metabolites in order to detect disease-related genotypes or mutations. Tests for metabolites fall within the definition of "genetic tests" when an excess or deficiency of the metabolites indicates the presence of a mutation or mutations. The conducting of metabolic tests by a department or agency that are not intended to reveal the presence of a mutation shall not be considered a violation of this order, regardless of the results of the tests. Test results revealing a mutation shall, however, be subject to the provisions of this order."
"GENETIC MONITORING

The term 'genetic monitoring' means the periodic examination of employees to evaluate acquired modifications to their genetic material, such as chromosomal damage or evidence of increased occurrence of mutations, that may have developed in the course of employment due to exposure to toxic substances in the workplace, in order to identify, evaluate, and respond to the effects of or control adverse environmental exposures in the workplace.

GENETIC TEST

(A) IN GENERAL - The term 'genetic test' means an analysis of human DNA, RNA, chromosomes, proteins, or metabolites, that detects genotypes, mutations, or chromosomal changes.

(B) EXCEPTIONS - The term 'genetic test' does not mean an analysis of proteins or metabolites that does not detect genotypes, mutations, or chromosomal changes."

"(5) GENETIC TEST- The term 'genetic test' means an analysis of human DNA, RNA, chromosomes, proteins, or metabolites, that detects genotypes, mutations, or chromosomal and biochemical changes.

(6) LABORATORY-DEVELOPED GENETIC TEST- The term 'laboratory-developed genetic test' means a genetic test that is designed, validated, conducted, and offered as a service by a clinical laboratory subject to CLIA using either commercially available analyte specific reagents (FDA-regulated) or reagents prepared by the laboratory (not FDA-regulated), or some combination thereof.

(7) PHARMACOGENETIC TEST- The term 'pharmacogenetic test' means a genetic test intended to identify individual variations in DNA sequence related to drug absorption and disposition (pharmacokinetics) or drug action (pharmacodynamics), including polymorphic variation in the genes that encode the functions of transporters, receptors, metabolizing enzymes, and other proteins.

(8) PHARMACOGENOMIC TEST

(A) IN GENERAL- The term 'pharmacogenomic test' means a genetic test intended to identify individual variations in single-nucleotide polymorphisms, haplotype markers, or alterations in gene expression or inactivation, that may be correlated with pharmacological function and therapeutic response.

(B) VARIATIONS AND ALTERATIONS- For purposes of this paragraph, the variations or alterations referred to in subparagraph (A) may be a pattern or profile of change, rather than a change in an individual marker."