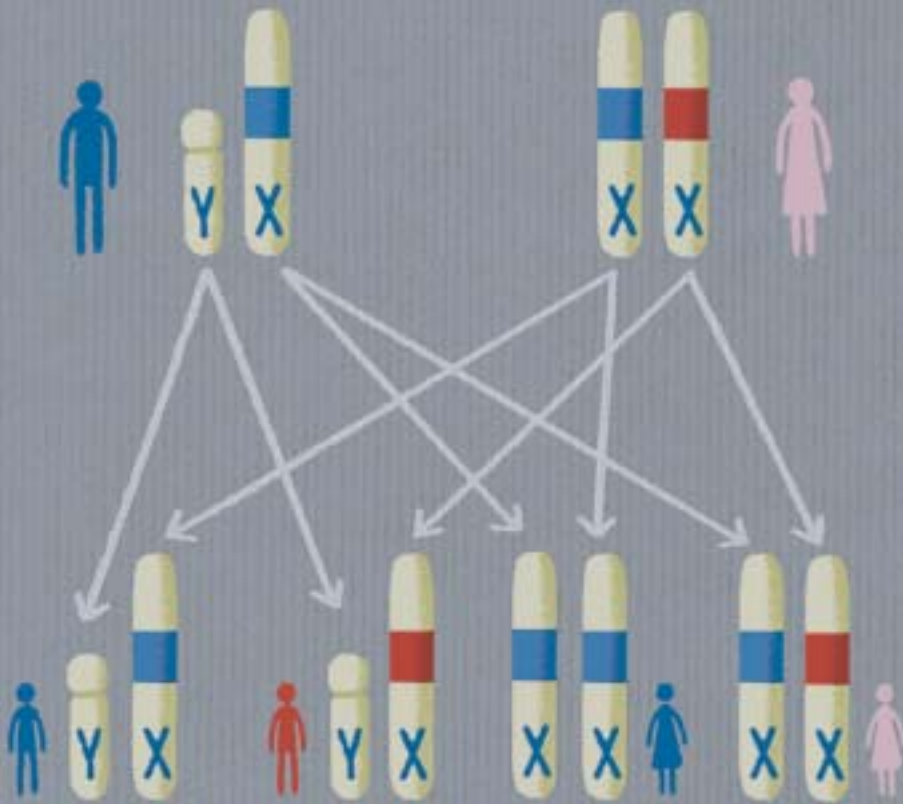




# 25 Recommendations on the ethical, legal and social implications of genetic testing



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# 25 Recommendations on the ethical, legal and social implications of genetic testing

by

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<b>Introduction</b>	<b>5</b>
The EC Expert Group	5
Working methods	6
The 25 recommendations	6
Preliminary remark	6

## The 25 recommendations

<b>General framework</b>	<b>7</b>
1. Need for universal standard definitions	8
2. Germinal and somatic genetic testing	8
3. “Genetic exceptionalism”	8
4. Public information and education	9
5. Public dialogue	10
<b>Implementation of genetic testing in healthcare systems</b>	<b>11</b>
6. Medical genetic testing and its context	12
7. Quality assurance	12
8. Population screening programmes	13
9. Genetic counselling	14
10. Data protection: confidentiality, privacy and autonomy	15
11. Protection from discrimination	15
12. Ethnicity and genetics	16
13. Gender issues and genetics	16
14. Social, cultural and economic consequences	17
15. Professional development	17
16. Partnerships and collaborations	18
17. Regulatory framework and criteria for test development and use	18
18. Rare diseases	19
19. Pharmacogenetics	19
<b>Genetic testing as a research tool</b>	<b>21</b>
20. Existing and new ‘biobanks’	22
21. Collections of human biological material and associated data and their uses	23
22. Cross-border exchange of samples	23
23. Informed consent	24
24. Samples from the deceased	25
25. Consent procedures for children and vulnerable individuals in human genetic research	25



# Introduction

When the news about deciphering the human genome, the so-called 'book of life', was announced, news channels predicted that this step in scientific analysis would make it possible to "banish inherited disorders, screen people for their vulnerability to diseases, tailor treatment to an individual's genetic make-up, create thousands of new drugs and extend human lifespan" (BBC, 27 June 2000). Today, just four years later, a range of genetic tests has been developed and the possibility of genetic testing is profoundly changing the methodologies and strategies used in medical therapy and healthcare. As these medical applications also encompass consideration of ethical, legal and social implications, careful reflection is needed to put an appropriate strategy into place to ensure the maximum potential of healthcare innovation.

At the moment, only a small group of informed people are familiar with the kind of decision-making and consequences these new technologies may infer. These people are familiar with genetic tests either because of their professional background or because they, or their families, have already been involved with such new and promising diagnostic tools as patients. Genetic testing will soon become part of everyday healthcare systems, and patients and professionals will have to learn to make decisions on the need for a test as well as understanding its consequences.

When integrating these technologies into the healthcare system, it is important to implement their application within a responsible framework of accompanying measures and activities. Trust and confidence should prevail when developing new testing capacities. In order to help decision-makers at all levels to introduce the necessary requirements rapidly, the European Commission's Research Directorate-General has invited a group of experts from various backgrounds to discuss the ethical, legal and social implications of genetic testing and to draw up relevant and urgently needed recommendations. What has to be considered when creating such a frame of responsibilities? What do decision-makers need to understand and to do now in order to give these new technologies a successful start as a powerful diagnostic and research and development tool?

## The EC Expert Group

The Expert Group invited by the European Commission to discuss the topic over the course of one year was multidisciplinary and included various stakeholders who were already involved or personally interested in the topic. Representatives came from the industry that produces or uses genetic tests, from NGOs (in particular, patient organisations with clear interests in the subject), and scientists and representatives from academic institutions with different backgrounds specialised in the field (law, philosophy, ethics, and medicine). The participants came from various national backgrounds within Europe and numbers were well balanced between men and women. Eryl McNally, Member of the European Parliament, acted as Chair of the Group and ensured that the experts achieved the necessary and agreed recommendations which should be useful for decision-makers in Europe. The discussion in the Group was both stimulating and encouraging.

## Working methods

Concentrating on genetic testing as a healthcare tool, the Group recognised the importance of a number of other areas which they did not address, such as issues regarding patenting, the use of genetic testing in insurance, judicial uses and forensic medicine (e.g. criminal, police), paternity testing, human gene therapy, fundamental research such as genome evolutionary studies, gene expression studies, and genomic and post-genomic applications, although all of them were addressed briefly.

The Group limited its discussion to issues and perceptions surrounding the application of genetic methodologies and technologies in health research, healthcare and related activities.

The ethical, social and legal criteria that played an important role when developing the following 25 recommendations were: reliability, quality and quality assurance, transparency, autonomy, education, respect of personal choice, information and consent, protection of vulnerable groups, protection of confidentiality, the right to know and not to know, the duty to disclose and the duty to warn in relation to liability, equity in access to healthcare, gender and minorities issues, control over human samples and data use in research and development, legal protection, and legal uncertainty.

## The 25 recommendations

The following 25 recommendations are organised into three chapters addressing:

- General framework;
- Implementation of genetic testing in healthcare systems; and
- Genetic testing as a research tool.

The 25 recommendations try, where possible, to address the relevant actors. Sometimes they also take the tone of a 'code of conduct'. Hence these recommendations should function partly as a 'code of conduct' for any actor in the field of genetic testing and partly as an 'action plan for genetic testing' to be implemented by policy-makers in the near future. The stakeholder conference, to be organised by the European Commission on 6-7 May, 2004 in Brussels, will discuss these recommendations further and will test whether or not they are understood, and which fields have yet to be adequately addressed.

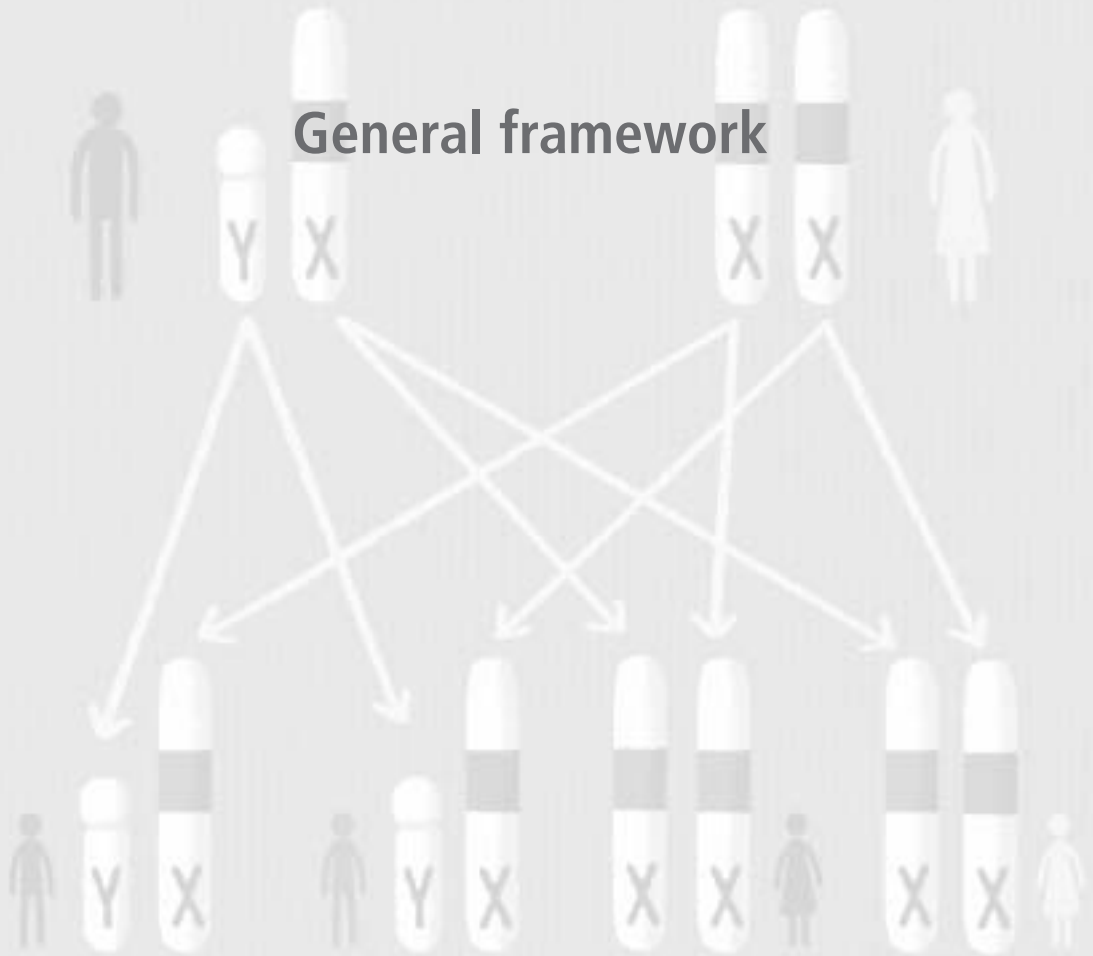
The Group considered its own discussions between the different parties as very fruitful and as a successful experiment in productive dialogue between various stakeholders. Consequently, social dialogue and its organisation have also been made a subject of the recommendations.

## Preliminary remark

The Expert Group shared the view that genetic testing represents progress in healthcare and an opportunity with the potential to lead to developments in preventive medicine. They recognised that there is no progress without cost and investment, and that all progress has both positive and negative implications. The conditions and context of the application of genetic testing therefore need assessment on a case-by-case basis to provide maximum benefit and minimum risk. Society needs to ensure that genetic testing is a matter of free choice and that it is never imposed.



# General framework



# 1. Need for universal standard definitions

There are various definitions of genetic testing and genetic data. Clarification of the issues at stake is an important prerequisite to any debate or official position. See report, introduction and Annex 2.

## Recommendation 1

That:

- a. any official statement or position should refer precisely to an explicit definition of the terms used or topic addressed;
- b. a consensus definition of genetic testing should be developed globally 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);
- c. the European Commission should consider taking the initiative on this topic.

The definitions used by the Group can be found in Annex 2 of the report. 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.

# 2. Germinal and somatic genetic testing

Further issues are at stake with respect to non-germinal (somatic) testing that require more in-depth reflection and investigation. See report, chapter 9.1.

## Recommendation 2

That:

- a. a specific working group be set up to discuss further issues relevant to genetic testing for acquired genetic properties.

# 3. “Genetic exceptionalism”

The sentiment that genetic data are different from other medical information (“genetic

exceptionalism”) is inappropriate. Genetic information is part of the entire spectrum of all health information and does not represent a separate category as such. All medical data, including genetic data, must be afforded equally high standards of quality and confidentiality at all times.

However, the current public perception that genetic information is somehow different is acknowledged by the Group. This perception is due to a number of factors. These include historical reasons (eugenics), the current predominance of predictive genetic tests for rare monogenic diseases which may give rise to particularly sensitive information affecting patients’ relatives, the fact that no treatment is available yet for most monogenic diseases, potential loss of control over samples, plus a number of other reasons.

Current efforts to establish guidelines, recommendations, rules, regulatory texts and laws that apply specifically to genetic testing and data handling should be viewed as an understandable response to specific public concerns. They are, however, only acceptable as a stepping stone to more considered and inclusive legal and regulatory frameworks that encompass **all** medical data and testing, and which reflect advancements made in healthcare provision. See report, chapter 3.2.

### Recommendation 3

That:

- a. “genetic exceptionalism” should be avoided, internationally, in the context of the EU and at the level of its Member States. However, the public perception that genetic testing is different needs to be acknowledged and addressed;
- b. all medical data, including genetic data, must satisfy equally high standards of quality and confidentiality;
- c. in order to track the evolution of public perception of genetic testing and to identify issues for future debate:
  - further research on ethical and social perceptions of genetic testing is necessary and should be promoted by the European Commission and national bodies; and
  - questions relevant to genetic testing should be included in pan-European surveys such as the Eurobarometer.

## 4. Public information and education

The new knowledge has to be disseminated quickly. As the scientific background is complex and not always easy to understand, it is imperative that opportunities for education are organised for both the public and the media. It is important to increase public awareness, education and understanding of genetic concepts. Genetic research and its clinical applications should be represented in an impartial way, and realistic expectations as to what they can achieve should be set. See report, chapters 3.1.2 and 7.2.1.

## Recommendation 4

That:

- a. materials and resources be developed and made available at the EU, national, and local level to provide information about genetic testing, genetic screening, and pharmacogenetics through a variety of media;
- b. science curricula at all levels (from primary to university level and vocational training) include reference to progress and potential in the field of medical genetics;
- c. national education systems ensure an adequate supply of appropriately trained scientists and teachers, including technicians and clinicians, to ensure that benefits arising from genetic research and genetic testing can be made real and delivered to all EU citizens;
- d. concerted efforts to promote dialogue, education, information and debate be encouraged;
- e. the 'Science and Society' component of the EC research and development framework be strengthened further.

## 5. Public dialogue

Dialogue should have a defined scope, be multidisciplinary, organised transparently and carefully designed to involve all relevant and interested stakeholders in an egalitarian fashion. Participants in the dialogue should be encouraged to be open-minded, willing to listen, respectful of local cultural values, and should treat this dialogue as an exchange of opinions rather than as an opportunity for proselytising.

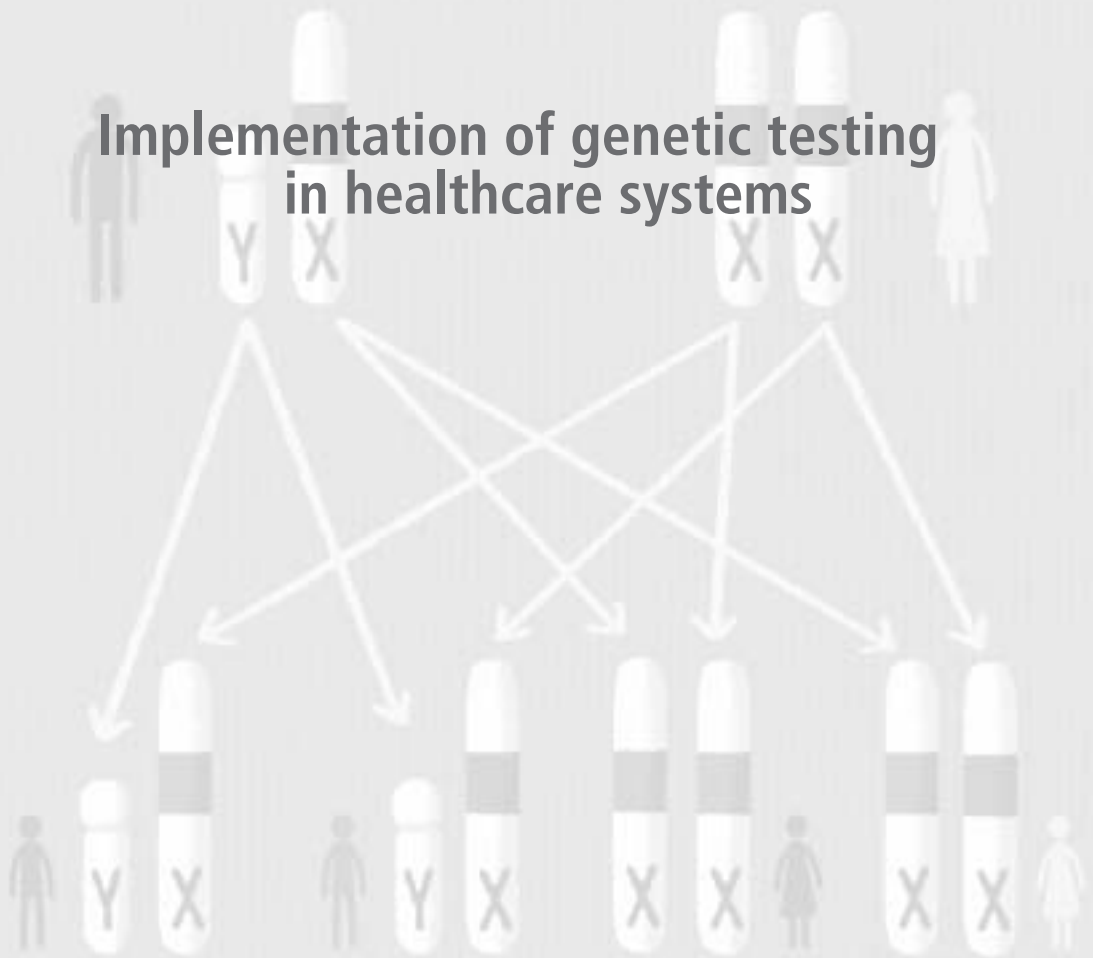
An effective dialogue will require discussion leaders to ensure that all participants in the debate are given equal opportunities to voice their respective positions, and that provision is made for questions and answers. See report, chapters 3.1.2 and 9.1.

## Recommendation 5

That:

- a. opportunities for public dialogue between different stakeholders be organised, offering participants equal opportunities for expression;
- b. different formats of dialogue and debate be organised as no single format will fit all purposes and all publics.

# Implementation of genetic testing in healthcare systems



## 6. Medical genetic testing and its context

Provision of genetic testing in the context of human healthcare requires appropriate procedures for obtaining informed consent, equitable access to testing as well as to appropriate counselling, and protection of confidentiality and privacy. These requirements will ensure that genetic testing offers new possibilities for individual choice rather than generating further constraints. The Group acknowledges the present activity of the Council of Europe<sup>1</sup> on such matters.

Predictive genetic testing has real potential to provide options for personal choice. However, it is imperative to recognise both the right to know and the right not to know as important individual rights. Equitable access to information and to quality genetic services must be assured. In addition, measures must be taken to prevent undesirable societal consequences as a result of genetic testing. The application of genetic testing for non-medical reasons requires careful consideration with regard to its potential consequences for society. See report, chapters 1.2, 3.1, 7.1.2 and 8.1.

### Recommendation 6

That:

- a. medically relevant genetic testing be considered an integral part of health service provision;
- b. medically relevant genetic testing should never be imposed and should always be a matter of free personal choice;
- c. comprehensive information about the availability of genetic tests be freely available from a range of reputable sources including public authorities, physicians, and patient groups;
- d. national healthcare systems ensure that genetic testing will be accessible equitably to all who need it.

## 7. Quality assurance

Genetic-testing services in Europe, while based on high-quality scientific know-how, tend to suffer from an intolerably high level of technical error and poor reporting. This is caused by a lack of both structuring and complementarity at European level and by the absence of a common European objective to provide quality services to all its citizens now and in the future. Diverse and heterogeneous quality schemes, a lack of reference systems, and differing Member State regulations, have added to the overall fragmentation of services. Nevertheless, genetic services face an ever-increasing number of requests for testing, while widespread susceptibility testing and pharmacogenetic tests are lurking on the horizon.

<sup>1</sup> <http://conventions.coe.int>

In the context of human healthcare, a test should only be offered when it has been shown to perform reliably and when there is a sound medical reason to consider it. A system for the validation of tests should be established. See report, chapters 1.4, 7.1.2 and 8.2.

## Recommendation 7

That:

- a. the European Union institutes a consistent regulatory framework to assure specific standards of quality for all genetic-testing services and their providers, including a system of accreditation for genetic-testing laboratories;
- b. test providers ensure that the information provided is accurate, by conforming with internationally agreed quality standards;
- c. national healthcare systems establish consistent quality requirements for genetic testing.

## 8. Population screening programmes

Genetic screening for increased disease risk will progressively become possible for common disorders. Offering screening to specific subgroups of the population for certain disorders may be beneficial, but may also carry risks. There must be agreement between medical professionals, patients, and the community that the benefits outweigh the risks. There is a need for meaningful information and regulation with respect to the provision of genetic screening on a population-wide level as well as the testing of certain subgroups within that population. See report, chapter 8.4.

## Recommendation 8

That:

- a. measures be put in place to ensure that tests are meaningful: the condition screened for must be serious, the test highly predictive, and follow-up actions must be available in terms of healthcare interventions (including reproductive choices);
- b. the relevance of the genetic condition being screened for be validated and regularly evaluated within the framework of the public health context (this may differ from country to country in the EU);
- c. the appropriate medical environment for providing information prior to testing and relevant post-test counselling be in place prior to offering such screening;
- d. pilot programmes be performed prior to general introduction of the screening;
- e. the economic dimension of envisaged screening programmes should be considered carefully.

## 9. Genetic counselling

The provision of specialised genetic counselling is considered an essential requirement for certain genetic tests, particularly for highly predictive tests for serious disorders. Provision of such counselling requires professionals who have received specific training. In other instances, the physician or other non-specialised members of the healthcare team may provide the patient with the relevant information. The provision of simple, printed information that can be consulted by the individual after counselling has been shown to be extremely valuable, so such materials should always be available.

Non-directive counselling must be the rule. The main goal of genetic counselling is to help individuals or families understand or cope with genetic disease, not to decrease the incidence of genetic disease. Appropriate explanations and sufficient time should be allocated to people in need of support. General information about genetic testing and counselling is part of regular medical practice and relevant education must be provided to physicians. Specialised counselling should be devoted to individual situations where it is of particular relevance.

The Group acknowledges, however, that while there should be a common basis of shared principles regarding standards of genetic counselling, practices will vary depending on cultural settings and values and therefore a certain heterogeneity of practices will continue to exist throughout the EU. See report, chapter 7.1.5.

### Recommendation 9

That:

- a. in the context of healthcare, genetic testing be accompanied by the provision of key information and, where appropriate, by the offer of individualised counselling and medical advice (in the case of highly predictive genetic tests for serious disorders, the offer of specific counselling should be mandatory, and patients should be strongly encouraged to take advantage of it);
- b. specific educational programmes on counselling and exchange of experience in the field be organised at the European level;
- c. specific qualifications and quality standards for those engaged in the provision of specific genetic counselling, whether clinicians or non-clinicians, be established and made mandatory;
- d. appropriate financial means for such training and the subsequent accreditation be made available;
- e. Europe-wide general standards for fundamental principles of genetic counselling be developed by relevant medical professional groups, with due consideration given to patients' views.



## 10. Data protection: confidentiality, privacy and autonomy

Public concern regarding genetic testing is grounded to some extent in the fear of misuse of genetic data and of inappropriate access to such data by third parties. Confidentiality and privacy with regard to all personal medical data, including those derived from genetic testing, are basic rights and must be respected. An individual's right to determine the extent of privacy and confidentiality that he or she will be afforded encompasses both access to personal information and data and to tissue samples as carriers of information. The individual has both the right to know and not to know. Affording legal protection in this field is of paramount importance and the existing EU Data Protection Directive<sup>2</sup> provides an appropriate framework for the need to protect personal data. See report, chapters 3.2, 4.1 and 7.2.2.

### Recommendation 10

That:

- a. genetic data of importance in a clinical and/or family context should receive the same level of protection as other comparably sensitive medical data;
- b. the relevance for other family members has to be addressed;
- c. the importance of a patient's right to know or not to know be recognised and mechanisms incorporated into professional practice that respect this. In the context of genetic testing, encompassing information provision, counselling, informed consent procedures, and communication of test results, practices should be established to meet this need;
- d. these issues are of particular relevance to vulnerable populations, whether in the EU or elsewhere in the world.

## 11. Protection from discrimination

Irrespective of confidentiality issues, personal medical data including genetic data must not be used to discriminate unfairly against individuals. The Group acknowledges the risk of discrimination as a result of genetic testing but notes that the field of discrimination extends far beyond genetics. Such discrimination may affect individuals, families or larger groups. Regarding prevention of discrimination in insurance and employment, the Group refers to the work of other bodies such as the European Group on Ethics in Science and New Technologies<sup>3</sup>, the Council of Europe<sup>4</sup>, and the European Society of Human Genetics<sup>5</sup>. See report, chapters 3.2 and 3.3.

<sup>2</sup> Directive 95/46/EC

<sup>3</sup> [http://europa.eu.int/comm/european\\_group\\_ethics/index\\_en.htm](http://europa.eu.int/comm/european_group_ethics/index_en.htm)

<sup>4</sup> See footnote 1

<sup>5</sup> <http://www.eshg.org/>

## Recommendation 11

That:

- a. data derived from genetic sources should not be used in ways that disadvantage or discriminate unfairly against individuals, families or groups in either clinical or non-clinical contexts, including employment, insurance, access to social integration, and opportunities for general well-being;
- b. EU-level regulations addressing these issues should be promoted;
- c. timely access to genetic testing should be based on need and appropriately resourced with no discrimination based on gender, ethnic origin, social or economic status.

## 12. Ethnicity and genetics

There may be differentiation of patient populations, based on ethnic groupings, with respect to genetic testing. Some genetic variants are more common in certain populations or groups in the EU and elsewhere. Specific attention should be paid to such groups in the context of test development and in the conditions set up for the use of these tests, both to ensure fair access and to avoid stigmatisation or stereotyping. Specifically, genetic tests are inappropriate to determine ethnicity and must never be used for this purpose. See report, chapter 3.3.2.

## Recommendation 12

That:

- a. genetic tests be clinically evaluated in the populations in which they are to be used;
- b. those who are involved in genetic research, the provision of genetic testing and healthcare policy-making be sensitive to the risks of stereotyping and stigmatisation based on ethnic origin, and recognise and respect ethnic and cultural sensitivities;
- c. minority ethnic groups should not be excluded from access to those genetic tests appropriate for them.

## 13. Gender issues and genetics

There are well-established differences between men and women regarding the risk of certain disorders, including genetic diseases. The Group considers that specific gender issues may arise in the context of genetic testing. However, little evidence for this exists so far. See report, chapter 3.3.1.

## Recommendation 13

That:

- a. further studies at EU level address the impact of genetic testing, in particular in societies where women and men are given different rights or privileges;
- b. governments and society be aware of the possible consequences of the application of genetic testing to aid reproductive choice for prospective sex selection;
- c. criteria be established at EU level to ensure that no gender discrimination occurs in the course of, or as a result of, EU-funded research projects.

## 14. Social, cultural and economic consequences

Relatively little information is presently available at EU level on the impact of genetic testing on health systems and on health economics. See report, chapters 8.3 and 8.5.

### Recommendation 14

That:

- a. the European Commission funds more research relating to the impact of genetic testing on the social, cultural and economic aspects of healthcare provision.

## 15. Professional development

The responsibilities of primary and specialised care providers will expand as genetic testing, including pharmacogenetics, evolves. This will require appropriate changes in medical school curricula. Other healthcare professionals will also require enhanced initial professional education and continuing professional development in the area of genetics. See report, chapters 7.1.3 and 8.1.

### Recommendation 15

That:

- a. initial educational and professional requirements be coordinated in all countries of the European Union;
- b. continued professional training be offered for healthcare professionals.

## 16. Partnerships and collaborations

The Group considers that all stakeholders, including governmental authorities, scientists, healthcare providers, industry and patient organisations should work together in a partnership approach to optimise future advances in healthcare that may become possible as a result of genetic testing, such as new treatment options and disease prevention. For effective development of new genetic tests and diagnostics, a productive interchange between academic scientists and those working in the healthcare industry will continue to be essential. See report, chapter 2.2.

### Recommendation 16

That:

- a. the European Union stimulates and supports partnerships between stakeholders;
- b. a framework for transparent collaboration between industry and academic scientists be established.

## 17. Regulatory framework and criteria for test development and use

The Group considers that a more explicit regulatory framework for test development needs to be set up throughout the EU. See report, chapter 6.2.2.

### Recommendation 17

That:

- a. the regulatory framework for genetic testing be further developed by the EU and other international organisations in a way that recognises both the need for new tests and the importance of safety, clinical validity and reliability;
- b. all newly developed tests must conform to the standards established before introduction into clinical use, based on a review process by an organisation or body independent of the test developer to ensure that the patient will benefit from the test;
- c. priority-setting for the development of accurate genetic tests be guided by the degree of unmet medical need, independently of disease prevalence;
- d. the EC takes measures to facilitate the availability of genetic testing for rare diseases as well as for more common diseases;
- e. the EC actively promotes the regulatory framework on these topics.

## 18. Rare diseases

The Group acknowledges that few countries have instituted screening programmes for serious rare diseases. See report, chapters 1.4 and 6.2.3.

### Recommendation 18

That:

- a. an EU-wide network for diagnostic testing of rare genetic diseases be created and financially supported as a matter of urgency;
- b. an EU-level incentive system for the systematic development of genetic tests for rare diseases be created and financially supported;
- c. for rare but serious diseases for which treatment is available, Member States introduce universal neonatal screening as a priority.

## 19. Pharmacogenetics

The term 'pharmacogenetics' describes the study of differences between individuals in drug response (efficacy or adverse reactions) which result directly from variations in DNA sequences and in differences in protein sequence, structure, or gene expression that are the direct consequence of such DNA variations. The main aim of pharmacogenetics is to help deliver medicines to patients who are most likely to benefit and least likely to experience adverse reactions. Although pharmacogenetics is currently still in a mainly exploratory phase, an increase in its application in healthcare is expected and appropriate measures should be prepared in time for this evolution. See report, chapter 5.

### Recommendation 19

That:

- a. national health authorities play a more active part in encouraging development of the field of pharmacogenetics:
  - by providing particular incentives to enable the development of pharmacogenetic tests and associated therapeutics which are clinically desirable but which may not be economically viable; and
  - by enhancing the possibilities of co-operation between industry, patients, and academia in this field;
- b. an appropriate harmonised legal, regulatory, and healthcare policy framework for pharmacogenetics be developed at EU level, taking into account research, therapy development, and clinical practice.



# Genetic testing as a research tool



A large part of research relevant to genetic testing relies on the use and exchange of human tissue samples and associated data because all biological samples contain DNA. These may allow, to a variable degree, identification of the individuals who are the source of the samples and data. Genetic studies also rely on genealogical, population, clinical, and personal data. The Group has concentrated on research aspects in terms of sample and data resources of human origin as indispensable means for the identification, validation, and development of genetic testing for potential clinical use, including pharmacogenetics, and on the relevant ethical, legal, and societal issues.

## 20. Existing and new ‘biobanks’

The term ‘biobank’ is used variably to designate several types of collections of biological samples. The Group has considered biobank collections to mean the biological samples themselves, plus the related databases, allowing a certain level of accessibility, availability and exchange for scientific studies. Public trust in research surrounding genetic testing is largely dependent on how the use of samples and data in and from such biobanks is undertaken and communicated. This applies in particular to the areas of informed consent, storage, data protection and the degree of anonymity of samples, the communication of study results and, where appropriate, of individual test results. A harmonised approach to these issues at EU level is considered desirable. The Group is aware that the Council of Europe is developing an activity regarding storage and research use of biological material and related data.

Depending on their scope and context, official identification and registration of such biobanking activities, as well as assurance of their long-term financial sustainability, are desirable and may be required. There is a need for education about biobanking and for guidelines to assure quality control of both the collections and the ethical management of such repositories.

For the study and validation of the association of genes with specific diseases, including the development of diagnostic products, devices and other tools, access to well-characterised human tissue samples for research and development use is essential. See report, chapter 4.2.

### Recommendation 20

That:

- a. guidelines be developed and coordinated across the EU to ensure that the use of samples, including those from archival collections, is not unduly delayed or impeded, particularly if proper consideration of their level of identification has been taken into account;
- b. action be taken by Member States to ensure that approval by a competent review committee is obtained before research is undertaken;
- c. an inventory of existing biobanks across the EU be created, indicating standards and rules of access, to identify which of their contents may or may not be used for genetic studies;



- d. a system be implemented to evaluate and monitor the current usage of existing biobanks throughout the EU;
- e. the task force on 'biological resource centres' set up by the OECD be followed closely by the European Commission regarding development of standards;
- f. the European Commission closely follows this activity.

## 21. Collections of human biological material and associated data and their uses

It should be noted that biobanking issues extend far beyond genetic testing. Few Member States have a well-developed legal framework in this domain. There is currently no EU directive regulating the use of tissue and cell conservation for research purposes. See report, chapter 4.2.

### Recommendation 21

That:

- a. the European Commission follows closely relevant activities and developments of the Member States in this field and in the global context;
- b. action be taken at the EU level, in coordination with other initiatives, to follow and address regulatory issues related to collections of human biological material and associated data and their uses.

## 22. Cross-border exchange of samples

Cross-border exchange of samples and data is important to improve European co-operation in this field. Ethical questions, such as validity of informed consent, play an important role in this transfer. See report, chapter 1.4.

### Recommendation 22

That:

- a. the European Commission evaluates the need for, and the feasibility of, developing harmonised standards for the research use of human samples and associated data (including informed consent issues), taking into account relevant international conventions on cross-border exchange of samples.

## 23. Informed consent

Consent issues for samples from individuals and groups entering new collections may differ from those applicable to existing collections, where contact may not have been maintained with the source individuals over the years. Explicit written consent is the rule but may not always be feasible for future uses of the samples and/or data not envisaged at the time of collection. The issues at stake include respect for individual or community autonomy regarding their control over the use of samples, the distribution and flow of data and samples across national borders, and methods that are both compatible with the nature of the research yet respect human rights. Various existing positions and practices exist in the EU. Professionals working in human genetics have developed recommendations and several ethical bodies have taken positions.

Genetic data represent personal information which may or may not have individual medical relevance, as well as medical or cultural relevance at the community level. Biological samples and associated or derived genetic or medical information of any kind and of any origin must not be collected, stored or used without obtaining informed consent based on adequate process and procedure, including appropriate approval by relevant review bodies, and consideration of community-related aspects where appropriate, regardless of the purpose of the collection and the level of anonymity. Samples for genetic diversity studies should only be obtained in compliance with local or national ‘traditions’ and regulations or laws. The sources of samples and individual data and the relevant agreement regarding their use should be documented in all protocols and publications. The informed consent process and procedures used should be fully transparent with regard to the planned research, including policies on provision of test results to individuals and to the population, as well as to the handling of samples and to the rights of sample donors. The Group considers that views of both patients and patient organisations, and public debate about biobanks, in particular with regard to consent issues, should be explored further. See report, chapters 3.3.2, 4.1 and 7.1.4.

### Recommendation 23

**That:**

- a. the European Commission promotes opportunities for dialogue between stakeholders to support exchange of experience throughout Europe on issues of sample and data use for research, at the individual, family and population level;
- b. the European Commission funds multidisciplinary research into the ethical, legal and social issues related to informed consent procedures for human genetic research and other relevant areas essential for any evolving research in genetics.

## 24. Samples from the deceased

As regards samples and genetic data from the deceased, the Group considers that in the case of the overriding interest of one or more blood relatives, even in the absence of consent given before death, their use could be legitimate: absence of consent should not be considered as equivalent to non-consent. Furthermore, their use should be permitted, when made anonymous, for research, development of genetic tests, and for teaching purposes. See report, chapter 4.1.

### Recommendation 24

**That:**

- a. Member States take actions to promote the right of access to samples and data from a deceased person, in the case of overriding interest for blood relatives;
- b. Member States take actions to allow the use of anonymous samples from the deceased for the purposes of genetic research, development of new genetic tests, as well as for teaching purposes.

## 25. Consent procedures for children and vulnerable individuals in human genetic research

As regards the participation of children or vulnerable people in genetic studies, the principle of acting in their best interest applies and specific protection is the rule, as in any research on humans. An important issue raised by long-term conservation and use of data and samples obtained from children relates to the question of when assent or consent, respectively, need to be obtained. The Group recognises that few data are available on “what children think” and how information should best be presented to them. See report, chapter 7.1.2.

### Recommendation 25

**That:**

- a. the use of tissue and accompanying data from minors or vulnerable individuals in research be permitted if, in so doing, their interests are served;
- b. specific consideration be given to children’s views, the information provided to them, and issues of children’s assent and/or consent.



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# 25 recommendations on the ethical, legal and social implications of genetic testing

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## General framework

1. Need for universal standard definitions
2. Germinal and somatic genetic testing
3. “Genetic exceptionalism”
4. Public information and education
5. Public dialogue

## Implementation of genetic testing in healthcare systems

6. Medical genetic testing and its context
7. Quality assurance
8. Population screening programmes
9. Genetic counselling
10. Data protection: confidentiality, privacy and autonomy
11. Protection from discrimination
12. Ethnicity and genetics
13. Gender issues and genetics
14. Social, cultural and economic consequences

15. Professional development
16. Partnerships and collaborations
17. Regulatory framework and criteria for test development and use
18. Rare diseases
19. Pharmacogenetics

## Genetic testing as a research tool

20. Existing and new ‘biobanks’
21. Collections of human biological material and associated data and their uses
22. Cross-border exchange of samples
23. Informed consent
24. Samples from the deceased
25. Consent procedures for children and vulnerable individuals in human genetic research