



EUROPEAN COMMISSION
Directorate General: RESEARCH

Survey on national legislation and activities in the field of genetic testing in EU Member States

Edited by Line MATTHIESSEN-GUYADER

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**Directorate E
Biotechnology, Agriculture and Food**



EUROPEAN COMMISSION
RESEARCH DIRECTORATE-GENERAL

Directorate E - Biotechnology, agriculture and food
The Director

INTRODUCTION

Genetic testing, its scientific, ethical, legal and social implications, has continued to be subject to debate both nationally and internationally. Discussions on the need or in some cases review of existing legislations or preparation of new legislation have been initiated across Europe. OECD is also active in this field and is preparing guidelines on quality assurance of genetic testing.

The European Commission has in its 2nd progress report on “Life Sciences and Biotechnology – a Strategy for Europe”¹, published on 6 April 2004, highlighted that “the various activities undertaken regarding genetic testing at European and international level have indicated the need for a co-ordinated approach on this emerging field within the Commission services and with the Member States”.

The report identifies the following priorities for future activities to be undertaken by the European Commission and Member States:

- to engage in **EU-wide co-ordination of efforts** to ensure the highest quality of genetic testing in the EU and beyond;
- to establish an **EU- wide networking of national centres** for exchange of information regarding quality assurance of genetic testing including training activities, and an **EU- wide networking for genetic testing of rare diseases**.

Without any intention to interfere with Member States’ competence regarding genetic testing, DG Research has established an informal working group involving officials and experts from Member States to ensure exchange of information and to identify actions which should be addressed at EU level in order to assure the highest quality of genetic testing. The need for collaboration and exchange of information at EU level was confirmed at the two meetings organised so far in May 2004 and March 2005.

This survey has been prepared with the help of the members of the informal working group, and I would like to thank sincerely the contact persons in each country for providing this information.

We would like to invite you to provide any further information you may have to Dr. Line Matthiessen-Guyader (line-gertrud.matthiessen-guyader@cec.eu.int).

Christian PATERMANN
Director

¹ COM(2002)27 final, COM(2004)250 final.

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Applications like medical registers and bio-banks are not directly addressed, but there is a framework of regulations on collection, storage and use of genetic data protecting privacy of applicants without jeopardising technical development.

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)
- Use of genetic testing and genetic information in employment (*ban*).
- Use of genetic testing and genetic information in health insurance (*ban*).

The Austrian Sicherheitspolizeigesetz, BGBl. Nr. 566/1991 and BGBl. Nr. 151/2004, is the legal framework for:

- Forensic use of genetic testing in criminal investigation and public security(permitted in some specific cases; restrictive)

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

A slight amendment of the Austrian Gentechnology Act taking into account recent medical progress is projected in 2005.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

No national reference centres and/or national expert laboratories have been selected thus far.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

A list of laboratories licensed to carry out predictive genetic testing can be found under the following URL:

<http://www.bmgf.gv.at/cms/site/attachments/8/9/5/CH0256/CMS1087982873584/>

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

There is no national network; individual labs collaborate at EU / international level.

Questionnaire on genetic testing and use of genetic information

COUNTRY: BELGIUM

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

The King Boudewijn Foundation Organized in 2003 as a citizen's conference. Contact www.mijngenen.be or tel 02 5111840 Recommendations are available from the website.

3. **Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.**

Royal decrees determine how the services can be provided. The centres that can provide genetic services have been identified in a Royal Decree and a specific Royal decree exists for forensic DNA testing. An additional Royal decree has set out the conditions under which Centres for Molecular Diagnosis (a joint venture between genetic centres, clinical chemistry, microbiology and pathology mainly for somatic diseases) can function. Other Royal decrees prohibit the use of genetic information by insurers and by employers.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

Not compulsory, but available for those who want to be accredited in addition to the European EQAs offered by EMQN and the CF Network. For Belgium the agency is Beltest, member of the European Accreditation agencies (EA). Contact person Jean Claude Libeer http://belac.fgov.be/beltest/home_en.htm. For EMQN the contact is Rob Elles: rob.elles@CMMC.nhs.uk, for the CF network the contact is Els.Dequeker@med.kuleuven.ac.be

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

Royal decree limiting genetic testing to the 8 Belgian genetic centers

With respect to clinical trials in Belgium, the EU directive has been transposed into Royal Decree published 2004-07-02.

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks?

- Use of genetic testing and genetic information in employment and obligatory public health insurance

A Royal decree prohibits the use of genetic information for employment or public health insurance.

- Use of genetic testing and genetic information in private life and/or health insurance

- *A Royal decree prohibits the use of genetic information for private insurance*

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

Royal decree fixing the conditions under which DNA testing can be ordered by the magistrate and Ministerial decree fixing the conditions for recognition of the labs.

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

A new law is in preparation regarding the applications of prenatal and preimplantation genetic diagnosis. Further, a revision of the tariffs for health insurance reimbursement of genetic tests and a law on the compulsory licensing of patented diagnostic tests are in preparation.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

No expert or reference centres have been identified officially for genetic testing. In practice however, some centres have specialised in testing for particular diseases e.g. connective tissue disorders, PGD.

For forensic testing the laboratories have been officially identified by the ministry mainly based on their accreditation under ISO 17025 and some other conditions.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

Yes. The Belgian centres participate in the Orphan platform project on rare diseases and are listed in ORPHANET. A separate website exists for the Centres for Molecular Diagnosis

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Networking exists as indicated above for a few diseases in the country, while samples are regularly send abroad when the service is not available within the country.

Questionnaire on genetic testing and use of genetic information

COUNTRY: CZECH REPUBLIC

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

Under the auspices of the Czech Molecular Genetics Society /www.slg.cz)

3. **Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.**

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

None

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

None

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

None

- Use of genetic testing and genetic information in employment and obligatory public health insurance

None

- Use of genetic testing and genetic information in private life and/or health insurance

None

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

None

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

Thus, a new law is being drafted by Social Democratic Legislators – but no details are available.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

*See COGENE report and
http://www.uhkt.cz/lab_a_vysetreni/nr_lab_dna_diag/dna_lab_db_en*

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

Yes - http://www.uhkt.cz/lab_a_vysetreni/nr_lab_dna_diag/dna_lab_db_en

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Yes - http://www.uhkt.cz/lab_a_vysetreni/nr_lab_dna_diag/dna_lab_db_en

Questionnaire on genetic testing and use of genetic information

COUNTRY: DENMARK

General comment: the in vitro directive is implemented in Danish law. Since the questionnaire apparently does not address commercial use of gene tests outside the healthcare and research context, directive-related issues are not detailed in what follows.

- 1. Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

Report from the Ad hoc Committee on Gene Technology, which was set up in consequence of a debate in the Danish Parliament in January 2001.

Principal recommendation: the capacity for genetic counselling should match future demands. The report (in Danish with an English summary) may be obtained from www.vtu.dk

- 2. Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

A lay person consensus conference (centred around a presumed representative group of lay persons) organised by the Technology Council in 2000 showed positive expectations in general. However also warned on possibilities of stigmatisation/discrimination on genetic grounds.

3. Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

Within the healthcare system, genetic tests and testing services are treated no otherwise than other kinds of tests/testing services. Labs are run under the responsibility of specialists (e.g. in clinical chemistry).

All the ca. 20 Danish labs performing genetic tests perform routine internal control procedures. Increasingly, they also participate in external quality control schemes (EQAS).

Also, a nationwide initiative on the establishment of quality indicators in health care is under way.

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

In those contexts, DNA-related testing and information are not treated principally otherwise than other tests and other kinds of sensitive personal information.

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

- Use of genetic testing and genetic information in employment and obligatory public health insurance

The employment-context covered by legislation 1996 (Law on the use of health related information). The employer may not ask for or use the results of predictive genetic tests. However, if the work situation poses a known risk to people with a definite genetic disposition, the employer may offer such testing, but the result of the test may only be revealed to the person (not to the employer).

- Use of genetic testing and genetic information in private life and/or health insurance

The health insurance context covered by legislation 1997 (Amendment to Law on insurance agreements)

Insurance companies may not ask for, receive or utilize the results of a predictive genetic test. Note: this does not include "family history".

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

Legislation concerning a register of dna-profiles for the use of identification purposes.

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

Further legislation is not under actual consideration.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

There are no officially appointed reference centres etc.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

No single database exists.

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

There is a close collaboration between the clinical genetics centres and collaboration within EU.

Questionnaire on genetic testing and use of genetic information

COUNTRY: ESTONIA

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes X no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

X yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

Active public discussion is going on in respect of Estonian biobank, but not directly on genetic testing as a diagnostic approach.

3. **Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.**

In Estonia there is only one DNA testing laboratory for inherited diseases. Few more for detecting microbial and viral pathogens and HLA testing.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

DNA testing laboratory at University of Tartu Clinicum is called Molecular Diagnostic Centre because this lab performs also the newborn screening program for all 14 000 newborns for PKU and HT. MDC is involved in QC system provided by EMQN

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)
-

Newborn screening is regulated on Ministry of Social Affairs level, DNA testing on University of Tartu Clinicum level, other is not taking place.

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and biobanks

Estonian biobank is regulated by the Human Genes Research Act approved by Estonian Riigikogu (Parliament) on year 2000. This is special law for the biobank. (www.geenivaramu.ee)

- Use of genetic testing and genetic information in employment and obligatory public health insurance

This is forbidden by the EGRAct.

- Use of genetic testing and genetic information in private life and/or health insurance

This is forbidden by the EGRAct.

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

For this Estonia has special DNA lab in Dept. of Police with special regulations

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

We have just one lab (MDC at the University of Tartu Clinicum) for human DNA testing (apart of the HLA typing for transplantations).

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

We do not have the database yet, however in the future we will have the database due to the EU Orphanet project: www.orphanet.ee; www.dnatest.med.ee or www.kliinikum.ee and then laboratoorium and then molekulaardiagnostika.

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Yes, it exists and we collaborate at international level. See relevant publications:

1. Walter S., Sandig K., Hinkel GK., Mitulla B., Öunap K., Sims G., Sitska M., Utermann B., Viertel P., Kalscheuer V., Bartsch O. Subtelomere FISH in 50 children with mental retardation and minor anomalies, identified by a checklist, detects 10 rearrangements including a de novo balanced translocation of chromosomes 17p13.3 and 20q13.33. AMERICAN JOURNAL OF MEDICAL GENETICS PART A 2004, 128A (4): 364-373
2. Öunap K., Uibo O., Zordania R., Kiho L., Ilus T., Öiglane-Shlik E., Bartsch O. Three patients with 9p deletions including DMRT1 and DMRT2: A girl with XY complement, bilateral ovotestes, and extreme growth retardation, and two XX females with normal pubertal development. AMERICAN JOURNAL OF MEDICAL GENETICS PART A 2004, 130A (4): 415-423
3. Salumets A., Horelli-Kuitunen N., Suikkari AM., Metspalu A., Tuuri T. Elevated incidence of chromosomally chaotic embryos among frozen-thawed preimplantation embryos. EUROPEAN JOURNAL OF OBSTETRICS GYNECOLOGY AND REPRODUCTIVE BIOLOGY 2004, 114 (1): 59-63

Questionnaire on genetic testing and use of genetic information

COUNTRY: FINLAND

- 1. Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

*Ministry of Health and Social Affairs organized a working group for Genetic testing in 1998. This working group has published a memorandum in 1998
The National Ethics Committee has given an opinion on the above memorandum in 1999
The National Ethics Committee has also given an opinion on the initiative for the Paternity law in 2001
No English versions available*

Contact person: Ritva.Halila@stm.fi

- 2. Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

*Public debate / discussions have not been organised.
Public seminars have been organised that have dealt with genetic testing and biobanks.
Contact person: Ritva.Halila@stm.fi*

- 3. Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.**

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

No legislation for quality assurance exists.

Licensing is needed for paternity testing. The Paternity testing laboratory of National Public Health Institute is accredited. Contact person for paternity testing: Anu.Jalanko@ktl.fi

Finland participates in OECD Working Party on Biotechnology; under this organization A quality assurance survey of 18 countries (incl. Finland) has been performed. Quality control available by EMQN.

Contact person for quality assurance: Mauri.Keinanen@labquality.fi

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

No specific legislation exists but some acts govern this area:

Convention on Human Rights and Biomedicine (Council of Europe)

Constitution of the Republic of Finland

Medical Research Act 488/1999

Personal data information act 523/1999

Act on the Status of Rights of Patients 785/1992

www.finlex.fi

Contact person: Mervi.Kattelus@stm.fi

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

No specific legislation exists but some acts govern questions related to genetic information and biobanks:

Constitution of the Republic of Finland

Medical Research Act 488/1999

Act on Medical Use of Human Organs and Tissues 101/2000

Personal data information act 523/1999

Act on the Status of Rights of Patients 785/1992

www.finlex.fi

Contact person: Mervi.Kattelus@stm.fi

- Use of genetic testing and genetic information in employment and obligatory public health insurance*

Act on the Protection of Privacy in Working Life (759/2004) specifically issues genetic testing in Section 15:

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

www.finlex.fi

Contact person: Matti.Lamberg@stm.fi

- Use of genetic testing and genetic information in private life and/or health insurance*

No legislation but an unofficial agreement states that genetic information is not used for health insurance purposes.

Contact person: Pekka.Koivisto@kela.fi

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

National Bureau of Investigation / Crime Laboratory performs DNA testing in criminal investigation.

This is governed by several Acts:

761/2003, 646/2003, 449/1987

www.finlex.fi

EC is well informed about the criminal investigations: Police collaboration work PCWG

Contact person: Matti.Karjalainen@krp.poliisi.fi

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

New legislation for paternity testing is under preparation in the Parliament.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

No official reference centres. Laboratory of Molecular Genetics at the Helsinki University Hospital may be considered one.

National coordinator of EDDNAL: Arto Orpana

Contact persons:

Arto.Orpana@hus.fi / Aarno.Palotie@helsinki.fi

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

No National Database of the 6 laboratories. Databases of tests do exist.

All the laboratories are in the international databases of EDDNAL and Gene.Test.org

The Mutation database of Finnish diseases has been collected to the www-pages.

www.Findis.org

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Finland is participating the EU Network of Excellence: EUROGENTEST: Contact person:

Helena.Kaariainen@utu.fi

Questionnaire on genetic testing and use of genetic information

COUNTRY: FRANCE

1. Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?

yes

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

The National Consultative Ethics Committee for Health and Life Sciences was established by a decree signed by the President of the French Republic on 23rd February 1983, and was enacted in the law of 29th July 1994. Now, according to the new law of 6th August 2004, the Committee's mission is to give opinions on ethical problems and societal issues raised by progress in the fields of biology, medicine, and health.

The National Consultative Ethics Committee is now an independent authority and is composed as follows: the President, nominated by the President of the Republic, an Honorary President, and 39 members. Five of these members are drawn from the main philosophies and religious faiths and are designated by the President of the Republic. Nineteen members are chosen because of their qualifications, competence, and their interest in ethical issues. Fifteen members are engaged in scientific research.

The National Consultative Ethics Committee makes the results of its work known in the following ways:

- *an annual public conference organised by the Committee on ethical problems in the field of health and life sciences*
- *press conferences which are organised when the Committee adopts an Opinion.*
- *a quarterly review called Les Cahiers du Comité. The Cahiers publish the full text of the Committee's opinions, recommendations and reports. Based on the theme of the most recent Opinion, a first chapter publishes official documents, experts' contributions, studies of regulations abroad, or original statements by personalities who are not members of the Committee. Another chapter concentrates on the Committee's recent activity and that of its members.*
- *via Internet, where all the Opinions and reports of the Committee are available.*

Président : Didier Sicard,

Adresse: 71 rue Saint Dominique 75 007 PARIS tel 33 01 44 42 48 52/53

mailto:contact@comite-ethique.fr

Six opinions of the CCNE are devoted to medical aspects of genetics. These opinions are in the six attached pdf files.

- Advice N°045 : Ethical questions arising from the transmission of scientific information concerning research in biology and medicine. (1995)
- Advice N°046 : Opinion and recommendations on "Genetics and medicine : from prediction to prevention". (1995)
- Advice N°076 : Regarding the obligation to disclose genetic information of concern to the family in the event of medical necessity. (2003)
- Advice N°076 : Ethical problems raised by the collected biological material and associated information data : "biobanks", "biolibraries". (2003)Advice
- Advice N°080 : Guidance of workers to risk-bearing occupations. Role of occupational physicians and reflections on the ambiguity of the concept of aptitude. (2003)
- Advice N°080 : Generalised prenatal screening for cystic fibrosis. (2004)

2. Will (or has) a public debate take (n) place on genetic testing and use of genetic information?

X yes

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

Two ways :

- Parliamentary debate for the Bioethics laws revision (August the 6th, 2004)
- Public debates organized by the National Consultative Ethics Committee (see above). Concerning genetic testing, meeting have been organized by this Committee in 1996, 2003 and 2004

3. Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate contact person(s).

Legal framework for:

** 3-1 Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)*

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The main texts of the legal framework are :

- The July 1994 Bioethics law, revised on August the 6th 2004 and all their application decrees.
- The In Vitro Diagnosis Medical Devices decrees.

The references of the texts framing genetic testing practice are :

- 16-10 to 16-13 from the Civil Code ;
- L. 122-45 from the employment code;
- L. 1131-1 to L. 1131-7, L. 1141-1 et L. 1141-2, L. 2131-1 to L. 2132-5 from the Public Health Code
- 225-1 à 225-4 et 226-25 à 226-30 from the penal code;
- 706-54 to 706-56 from the penal procedure code.

These texts are coming from the following laws :

- Law n° 94-653 July the 29th 1994 about human body respect ;
- Law n° 94-654 July the 29th 1994 about donation and use of human body elements (organs, cells,..) and products (blood,...), assisted procreation and prenatal diagnosis ;
- Law n° 2002-303 March the 4th 2002 about patient rights and health system quality (and notably its articles 4 and 98) ;
- Law n° 2004-800 August the 6 2004 about Bioethics ;
- Concerning the use of genetic tests and genetic data in the field of legal procedures: law n° 98-468 June the 17th, 1998 about sexual aggression and minors protection, law n° 2001-1062 November the 15th, 2001 about everyday security, law n° 2003-239 March the 18th, 2003 about the Interior security.

In order to perform genetic testing in a medical purpose the health establishments or the medical diagnosis laboratories have to obtain an authorization and the Biologists have to be licensed (the authorizations and licenses will be delivered by the Biomedicine Agency as soon as it will be created. At the present day they are delivered by the Prefect). Regulations are different for prenatal diagnosis and for genetic characteristics determination.

At the present day external quality assurance control is limited to classical biological analyses (the French National Agency for Sanitary Security of Health Products, AFSSAPS, is in charge of such controls). There is no external quality assurance controls for genetic tests. There is a project of external quality assurance control for cystic fibrosis genetic tests.

*** 3-2 Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)**

Genetic testing are performed as well for symptomatic as asymptomatic subjects (the regulations are different in the two cases) in the view of the diagnosis of their disease. They are also performed for a prenatal diagnosis. Pharmacogenetic tests are performed before the prescription of some drugs.

HLA typing in the view of a transplantation is excluded of the genetic testing regulations.

*** 3-3 Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio- banks**

Two laws (Bioethics and Public Health) promulgated in August 2004. Application decrees to be published probably next summer

*** 3-4 Use of genetic testing and genetic information in employment.**

See below

*** 3-5 Use of genetic testing and genetic information in obligatory public health insurance and private life and/or health insurance**

Concerning points 3-4 and 3-5 the legal framework is described in point 3-1.

A principle has to be especially underlined (article 16-13 of the Civil Code) « Nobody may be the object of any discrimination because of his genetic characteristics » a violation of this principle is liable to prosecution (articles 225-1 to 225-4 of the penal code).

In the field of employment the use to the genetic characteristics of a candidate is strictly forbidden, whatever the type of employment is, according to article L. 122-45 from the employment code.

In the field of insurance, the use to the genetic characteristics is strictly forbidden, even if they are transmitted by the customer according to article L. 1141-1 of the public health code.

*** 3-6 : Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)**

« The use of genetic fingerprint in the legal procedures is planned in articles 706-54 to 706-56 from the penal procedure code.

The magistrates (state prosecutor and investigating magistrate) and the official empowered, are allowed to order the search for genetic fingerprints in the scenes of crimes, as well as on people under sentence and introduce them in the national file of genetic fingerprints (fichier national automatisé des empreintes génétiques, FNAEG).

The March 18th 2003 law on the Interior security has established the infractions of the law for which the genetic fingerprint determination of a subject may be ordered (art. 706-55 of the penal procedure code) ; this list include notably terrorism actions and offences to the “fundamental interests of the nation».

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

The Bioethics laws have been revised few month ago (law n° 2004-800 August the 6th 2004). The application decrees should be published within few months.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

As it will be described in question 8, networks for genetic testing in rare diseases are progressively created since 2001. At the present day there is three main networks. Each one is managed by 4 reference laboratories. A lot of criteria are used to select the reference laboratories. Some examples of used criteria are : (1) the number of complex tests (prenatal diagnosis, complex mutations, screening for mutations in the whole gene, ...) performed every year (2) publications of the laboratory ; (3) the members of the laboratory must belong to a recognized research team ; (4) international reputation,... The reference laboratories are selected by an expert committee including up to 50% of non-French experts (European experts).

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

The list of the medical laboratories which are authorized to perform genetic testing is available upon request to the French Ministry of Health. The list is also available on the Orphanet web site (www.orpha.net). Within few months the Biomedicine Agency (it will be created on May the 10th 2005) will be in charge of this list and its updating. The Biomedicine Agency will be also in charge of the evaluation of the laboratories performing genetic tests

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

A first network of laboratories performing genetic tests for rare diseases have been created by the French Ministry of Health in 2001. It is devoted to cystic fibrosis. A second large network have been created in 2002. It is devoted to molecular genetics of rare cancers. A third large network for muscular dystrophies, mental retardation and neurogenetics have been created in 2003. A last large network, devoted to the genetic testing of all the others rare diseases should be created in 2005. At the present day these networks have no European or international collaborations.

Questionnaire on genetic testing and use of genetic information

COUNTRY: GERMANY

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

X yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

The German National Ethics Council, that has been set up by the chancellor in 2001 has provided an opinion on Biobanks for research and on Genetic diagnosis before and during pregnancy.

*Further information is available at the web address
www.ethikrat.org/_english/publications/opinions.html*

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

X yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

Bürgerkonferenz: Streitfall Gendiagnostik (April 2001) – a conference of randomly selected citizens representing the German population with the task to draft an opinion on genetic testing. The conference covered three main issues: genetic tests to diagnose and prevent diseases, preimplantation genetic diagnosis, and prenatal diagnosis. Their opinion emphasizes the need for public information, and states that genetic tests may be conducted only by qualified doctors. The conference recommends - among other things - the implementation of a central commission for certification of laboratories and genetic tests and addresses the potential for misuse of predictive genetic testing results, especially in employment and private health insurance. The conference participants expressed their opinion that genetic data belong solely to the person concerned and must be secured against third parties.

Contact: www.buergerkonferenz.de

The conference was part of a research project on ethical, legal and social issues (ELSI) with regard to human genetics and was funded by the Federal Ministry of Education and Research.

3. Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.

Currently, there are no specific regulations governing genetic testing and the use of genetic data.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

At least, generally a license (or approbation) is required to operate any laboratory providing medical tests. The approval lies within the responsibility of the medical self-governing body (i.e. associations of doctors, the German Hospital Federation and the health insurance funds) as is the case with guidelines regarding quality assurance requirements. There is no specific license required for genetic testing.

Accreditation is voluntary. Special criteria for accreditation of molecular genetic testing laboratories are in preparation. The German accrediting agencies work according to DIN-EN-ISO standards 15189 or 17025 for accreditation and 9001 for certification. However, existing criteria and check-lists for accreditation are oriented to the requirements of routine clinical laboratories.

*Contact: Prof. Dr. rer. nat. Müller-Reible, expert at the German Society of Humangenetics; Institut of Humangenetics, University Wuerzburg, Biozentrum, Am Hubland, D-97074 Wuerzburg, Tel: (+)49-931-888-4063, Fax: (+)49-931-888-4069
e-mail: crm@biozentrum.uni-wuerzburg.de*

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

No specific legislation exists relating to the use of genetic testing and genetic information in health care. For some genetic applications for example newborn screening directives exist set up by the Federal Joint Committee (Gemeinsamer Bundesausschuss), which regulates the provision of medical services that are covered by the statutory health insurances.

Contact: Gemeinsamer Bundesausschuss, Auf dem Seidenberg 3a, 53721 Siegburg, Tel.: +49-2241 - 9388-0, Fax: +49-2241 - 9388-573

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

In the absence of specific legal provisions, the use and protection of genetic information is subject to the provisions of the Federal Data Protection Act, in particular, and the data protection laws of the Federal Laender.

- Use of genetic testing and genetic information in employment and obligatory public health insurance

In this respect, there are no legal regulations.

- Use of genetic testing and genetic information in private life and/or health insurance

While private health insurance is not covered by legal regulations, the Gesamtverband der Deutschen Versicherungswirtschaft e.V. (German Insurance Association - GDV), has adopted a self-commitment under which insurance companies have committed themselves neither to make predictive genetic testing a prerequisite for securing a contract, nor to require the client to submit to the insurance company, prior to conclusion of the contract, any predictive genetic test carried out on a voluntary basis. In this respect, the insurance industry exempts prospective members from the duty to disclose risk-relevant conditions prior to concluding a contract, as stipulated in the German Insurance Contract Act. Excepted from this exemption shall be life assurance policies involving very high amounts.

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

In the forensic field, legal provisions are already in place, governing the permissibility of the so-called genetic fingerprint in investigating crime. Therefore, this field will be exempt from the scope of the Genetic Diagnosis Bill that is currently under preparation.

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

According to the Coalition Agreement concluded between the governing parties, it is intended to adopt the Genetic Diagnosis Bill later in this legislative period.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

No reference laboratories for human genetic testing exist in Germany.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

Yes. A database of genetic testing labs is created by the Berufsverband Deutscher Humangenetiker at <http://www.bvmedgen.de/welcome.html> (click on "Qualitätsmanagement/Diagnostik" and go to "QM-Datenbank"). Registration with the database is voluntary. The next version of the database will also be available in English and will contain additional information on the accreditation status and external quality assessment participation of labs.

A number of German laboratories are also listed at Orphanet (<http://www.orphanet.de/>) and/or EDDNAL (www.eddnal.com).

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

In 2003, the Federal Ministry of Education and Research has launched a 5-year program to establish networks on rare genetic diseases. Total funding volume is 25 Mio Euro. At present, 10 networks have been established. Details can be found at

http://www.gesundheitsforschung-bmbf.de/foerderung/Vorhabenuebersicht/1-Bekaempfung-Krankheiten/Seltene_Erkrankungen

Some of the networks are linked to EU projects with related topics (e.g. the German Skelettdysplasie Netzwerk - SKELNET to the European Skeletal Dysplasia Network (ESDN at www.esdn.org) or the German Muskeldystrophie-Netz to the EUROMEN project (http://www.myocluster.org/f_euro.html)).

Activities/developments in Germany relating to genetic diagnosis

Since the performing of genetic testing and the use of genetic information are not yet governed by specific legal regulations in Germany, it is intended – as laid down in the Coalition Agreement – to pass a comprehensive Genetic Diagnosis Act later in this legislative period.

The key regulatory content of this Genetic Diagnosis Act will include the protection of informational self-determination in the context of genetic testing and the use of any results obtained in the process. In addition, individual information and education, genetic counselling, the requirement of prior consent and the right not to know are to be standardised in this Act. The latter is also intended to include the relevant doctor's prerogative, quality assurance measures as well as provisions to ensure the protection of genetic information and genetic samples. Over and beyond that, the scope is to cover genetic testing for medical purposes, including prenatal genetic testing, for life-planning purposes, for paternity testing, for research purposes, in the context of private insurance and in the world of work, including employment contracts under public law. Last but not least, this piece of legislation will address the use of genetic information and genetic samples. Issues likely to be excluded from the Bill's scope are, *inter alia*, provisions on pre-implantation and polar body diagnosis, tests for acquired somatic mutations and the performance testing of medical devices.

The fact that questions relating to the admissibility of genetic testing are significant beyond the national domain, becomes obvious especially when looking at genetic testing in the context of employment relationships and the private insurance business, since it is possible for each EU citizen to both take out, for instance, a life insurance policy and enter into an employment relationship in any Member State of the Union. Consequently, this Ministry is actively co-operating, on the supranational level, in the development of international legal framework instruments on genetic medicine. These instruments include the UNESCO Declaration on the Protection of the Human Genome, the Additional Protocol on Human Genetics that will further develop the Council of Europe's Convention on Human Rights and Biomedicine and the OECD's Survey on Quality Assurance and Proficiency Testing Schemes.

The findings of this survey prove that the setting of uniform quality standards for genetic diagnosis is of major importance on the national level as well. Therefore, the Genetic Diagnosis Act is also planned to include provisions on quality assurance and accreditation. The Genetic Diagnosis Act will, therefore, lay down the requirements for genetic testing, protect the personality rights of the citizens and safeguard them from genetic discrimination. At present, the envisaged provisions are still under discussion. As soon as the relevant Bill has been tabled in the Bundestag, it will also be accessible to the public.

Questionnaire on genetic testing and use of genetic information

COUNTRY: IRELAND

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

From Dr. Siobhán O' Sullivan, Scientific Director, Irish Council for Bioethics www.bioethics.ie

The Irish Council for Bioethics have not provided an opinion on the use of genetic testing/ genetic information in research/health/care. We are however in the currently drafting a report (should be released in April) giving guidance on use of biological material in research. We do not differentiate between genetic /non-genetic research. We address quite comprehensively issues of data protection, disclosure and consent which would all be relevant in the context of genetic research. Other relevant documents include: The Irish College of General Practitioners have published a guidance document in 2004 entitled "Ethical Questions to be considered by a Research Ethics Committee when approving Clinical Trials which involve Genetic Testing". It is on the ICGP website but only available to members who have a login password.

The Health Research Board published a document in 2002 entitled "Genetic Research and Human Biological Samples". The Author is Asim Sheikh and the document is available on the HRB website www.hrb.ie

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

Again, from Dr O'Sullivan:

There has not been a wide-ranging public debate on genetic testing in Ireland as yet. The Council have talked about this issue on occasion in relation to genetic testing and insurance, employment in interviews with the print media and also on radio. However, this has been in the context of raising awareness of bioethical issues in the general sense.

3. **Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.**

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

None in existence or proposed

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

Currently none, but legislation proposed.

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

Only as covered by the Data Protection Act, no specific provisions for genetic information.

- Use of genetic testing and genetic information in employment and obligatory public health insurance

We have no obligatory health insurance. Proposed legislation would ban use of genetic information in employment.

- Use of genetic testing and genetic information in private life and/or health insurance

Currently a limited moratorium, but legislation proposed.

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

Specifically excluded from protection under proposed legislation (Disability Bill, see below).

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

Disability Bill published 2004 contains provisions regulating the use of genetic testing and genetic information. PDF of bill attached

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

National Centre for Medical Genetics established by DoHC grant in 1994, with Clinical Genetics, Cytogenetics and Molecular Genetics divisions. www.genetics.ie

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

Only an informal database held at National Centre for Medical Genetics

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Ireland joined Orphanet in 2004, network is being established. No rare disease testing activity outside the National Centre for Medical Genetics.

Extract from
Government of Ireland
Disability Bill, 2004

The full document is available at
<http://www.oireachtas.ie/viewdoc.asp?fn=/documents/bills28/bills/2004/3904/3904.pdf>

PART 4
GENETIC TESTING

This Part introduces safeguards for the use of genetic data in general, especially for employment purposes and outlaws its use for insurance purposes. These safeguards will facilitate access for people who may be affected by genetic disorders. The use of family history information for insurance purposes may be restricted by regulation. The provisions of this Part will be subject to review commencing no later than 1 January, 2014.

Section 39 is a definitional section. “Genetic data”; “genetic testing”; and “processing” are defined for the purposes of this Part. The definition of processing is the same as in the Data Protection Acts and this includes the actual obtaining of data.

Section 40 provides that genetic testing may only be carried out where the person who is the subject of the test has given consent for the use of the resulting data in accordance with the Data Protection Acts, unless the testing envisaged is prohibited by law. The section prohibits the processing of genetic data in relation to the employment of a person unless the processing has been approved beforehand by the Data Protection Commissioner. The section outlaws the processing of genetic data in relation to the assessment of a person for insurance, a pension product/arrangement, or a mortgage. Limitations are also placed on the processing of genetic data for other purposes by requiring the processor to take all reasonable steps to ensure the person who is the subject of the test, is aware of the purpose and possible outcomes and effects of the data processing. Failure to observe the provisions on processing genetic data will constitute an offence.

Section 41 also provides that information about the family history of an applicant for insurance must be processed in accordance with regulations which the Minister may make under the Data Protection Acts. Before making these regulations, and depending on which financial product the data processing pertains to, the Minister shall consult with either the Data Protection Commissioner, the Minister for Health and Children, the Minister for Social and Family Affairs or the Irish Financial Services Regulatory Authority.

Section 42 requires that the operation of this Part will be reviewed by the Minister not later than 2014. In carrying out the review, the Minister must consult with other relevant Ministers, the Data Protection Commissioner, the Irish Financial Services Regulatory Authority and any other relevant persons or bodies.

Section 43 confirms that nothing in this Part shall authorise the processing of personal data which would be contrary to the Data Protection Acts 1988 and 2003 or prohibit the use of genetic testing by the Garda Síochána for forensic or criminal investigation or other lawful purposes.

Extract from Explanatory Memorandum:

The full document is available at
<http://www.oireachtas.ie/viewdoc.asp?fn=/documents/bills28/bills/2004/3904/3904.pdf>

PART 4 GENETIC TESTING

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Section 43 confirms that nothing in this Part shall authorise the processing of personal data which would be contrary to the Data Protection Acts 1988 and 2003 or prohibit the use of genetic testing by the Garda Síochána for forensic or criminal investigation or other lawful purposes.

Extract from Guide to the Disability Bill, 2004 issued by the Department of Justice and Law Reform

The full document is available at

[http://www.justice.ie/80256E010039C5AF/vWeb/flJUSQ653ETZ-en/\\$File/Disability+Bill+Guide.doc](http://www.justice.ie/80256E010039C5AF/vWeb/flJUSQ653ETZ-en/$File/Disability+Bill+Guide.doc)

PART 4: GENETIC TESTING

Genetic testing (sections 39–43)

This Part provides safeguards for the use of information obtained from genetic testing. The provisions aim to ensure that people who may be affected by genetic disorders will not be subject to any unreasonable requirements from an employer or an insurance or mortgage provider. The protections provided are in addition to the substantial safeguards for the use of personal information contained in the Data Protection Acts. These new safeguards will be reviewed in 2014.

The safeguards provide that -

- genetic testing may only take place with a person's consent, in accordance with the Data Protection Acts
- the results of a genetic test can't be used in relation to insurance, a mortgage, a personal pension or employment
- the person being tested must be made aware of the intended use of the test results and must, as far as possible, be informed about the possible outcomes of the test
- the use of a person's family history for insurance purposes may be regulated by the Minister after consultation with other relevant Ministers, the Data Protection Commissioner and other interested bodies or groups.

Questionnaire on genetic testing and use of genetic information

COUNTRY: ITALY

1. Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?

X yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

*We have a document issued in 1999 by the Italian Bioethics Committee, "Orientamenti bioetici per i test genetici (19 novembre 1999)", SINTESI E RACCOMANDAZIONI SYNTHESIS AND RECOMMENDATIONS are available at: <http://www.palazzochigi.it/bioetica/testi/191199genetici.html>
The coordinator was: Prof. Alberto PIAZZA-University of Turin; alberto.piazza@molinette.unito.it.*

The document goes through important ethical objectives in: genetic tests and screenings, genetic counselling, predictive tests, genetic tests in oncology, tests performed on minors and children, genetic tests at the workplace, genetic test and patients with complex behaviour personalities, genetic data storage, genetic discrimination. Specific recommendations are made about: the Human Genome Project, genetic tests in oncology, genetic tests and employment, genetic tests and insurance, genetic discrimination.

The text is organised in four parts.

The first part focuses on:

- a) general scientific background information on DNA and biotechnologies*
- b) definition, peculiarity and different uses of genetic tests.*

Objectives ethically important in the context of genetic testing and genetic screenings are:

- To contribute to health improvement of people affected by genetic diseases and/or*
- To allow disease carriers to make reproductive choices on a basis of exhaustive information and/or*
- To contribute to alleviate family/community anxiety when dealing with serious genetic diseases*

The importance of genetic counselling is stressed as the major process involved in the communication of information and risks to the individual and/or the family.

The second part refers to the state of the art of Italian structures performing genetic testing (in 1999) and regulatory frameworks.

The third part focuses on bioethical implications of the complex issue "genetic testing".

Here we report some recommendations (not all):

- To avoid any form of discrimination depending on individuals/families genotype*
- To consider the complexity of ethical issues in predictive medicine, and specifically in the use of predictive genetic tests*
- To give to people the right to know and not to know the genotype, especially when there is no advantage. (i.e. for the therapy)*
- Predictive tests in oncology can be very important for prevention, but can cause psychological problems. These tests should be recommended*
 - i) to affected patients, when the genetic diagnosis can modify and/or predict the diagnosis/prognosis of the disease*
 - ii) to asymptomatic patients' relatives, to be included in follow-up programs for an early-diagnosis of expected tumours and/or to evaluate the access to prophylactic surgery*
 - iii) to an asymptomatic individual, when genetic diagnosis can lead to a change in the lifestyle and in the diet, or to a protection from risk factors.*
- In the workplace any form of discrimination should be avoided. When there is a possible risk of genetic damage to an employee resulting from some component of the working environment, the employer has the duty to eliminate that risk.*
- Health insurances should not take in account genetic information, especially about polygenic and multifactorial diseases.*
- Genetic screenings have to be approved by consensus by the International Scientific Community; criteria adopted to screen populations and the test reliability (sensitivity and specificity) must be known*
- Genetic data must be confidential; discrimination must be avoided.*

A chapter dedicated to European and International regulatory frameworks and a glossary is at the end of the text.

2. Will (or has) a public debate take (n) place on genetic testing and use of genetic information?

X yes No

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

A wide debate has taken place about the law 40/2004 concerning medically assisted procreation (<http://www.parlamento.it/parlam/leggi/04040l.htm>); however, the debate is not directly linked to genetic testing and use of genetic information.

A referendum to change the law has been proposed and will be performed by 2005 (see question 4).

3. Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.

In 1999 the document “Linee guida per test genetici Guidelines for genetic testing” was issued by the Comitato Nazionale per la Biosicurezza e le Biotecnologie (Presidenza del Consiglio dei Ministri) and the Istituto Superiore di Sanità (Public Italian Health Institute) (available at <http://www.cnmr.iss.it/TEST/DOCU/lgui.pdf>). It goes through relevant issues in genetic testing, including confidentiality, data protection and informed consent.

In 2004 the document “Linee guida per le attività di genetica medica “ has been issued (Conferenza Stato Regioni, Seduta del 15 Luglio 2004, Presidenza del Consiglio dei Ministri) (available at <http://www.cnmr.iss.it/TEST/DOCU/LGTG2004.pdf>). The text contains guidelines for medical genetics services.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

According to the document “Guidelines for medical genetics services” (Linee guida per servizi di genetica medica, Conferenza Stato Regioni 2004), Medical Genetics Services have to participate to internal and external quality assessment programs according procedures established by regional systems. External quality assessment (EQA) has to be performed at a regional, national and/or European level. The Italian National EQA is coordinated by the Istituto Superiore di Sanità (see p17 paragraph 8.2.3).

Up to now, the Italian National EQA involves 87 public diagnostic genetic laboratories (cytogenetics and molecular genetics) distributed on the national territory; it is coordinated by the Istituto Superiore di Sanità and is funded by the Italian Ministry of Health.

Organization and first year results of the EQA Scheme are described in the following paper: “Quality assessment in cytogenetics and molecular genetic testing: the experience of the Italian Project on Standardization and Quality Assurance”. D. Taruscio, V. Falbo, G. Floridia, M. Salvatore, C. Pescucci, A. Cantafora, C. Marongiu, A. Baroncini, E. Calzolari, A. Cao, G. Castaldo, F. Dagna Bricarelli, G. Guanti, L. Nitsch, P.F. Pignatti, C. Rosatelli, F. Salvatore, O. Zuffardi. Clin Chem Lab Med 2004;42(8):915-21.

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

a) The document “Conferenza Stato-Regioni” (Guidelines for Medical Genetics Services) describes the use of different genetic tests; a paragraph is dedicated to informed consent and ethical problems, including the issue of genetic counselling (paragraphs 3, 4, 7)

b) The data protection law N°D.L.196 (30 giugno 2003) (CODICE IN MATERIA DI PROTEZIONE DEI DATI PERSONALI in Gazzetta Ufficiale n. 174 del 29 luglio 2003 - Supplemento Ordinario n. 123, available at <http://www.parlamento.it/parlam/leggi/deleghe/03196dl.htm>) regulates data treatment in

health care (see artt 75-160). In particular, article 90 refers to genetic data; bone marrow donors must remain anonymous (comma 3 art. 90).

English version of the data protection Law N°D.L.196:
<http://www.garanteprivacy.it/garante/document?ID=727068>

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

Informed consent is required and data protection is regulated by the data protection law N°D.L.196, 30 giugno 2003) (CODICE IN MATERIA DI PROTEZIONE DEI DATI PERSONALI in Gazzetta Ufficiale n. 174 del 29 luglio 2003 - Supplemento Ordinario n. 123, available at <http://www.parlamento.it/parlam/leggi/deleghe/03196dl.htm>- Art.94 Banche di dati, registri e schedari in ambito sanitario)

International Data Transfers – The data protection law (DL 196, 2003) has incorporated and, to some extent, updated the previous rules on data transfers (data transfers are addressed in Sections 42-45 of the code). Whereas previously businesses had to notify the Garante of their intention to transfer data outside the EU, under the new system companies will only have to provide notification in cases in which the transfer of data could prejudice data subjects' rights (see the Notification section).

The rules for legitimising transfers to non-EU countries can be found in Section 43 of the code and include consent, meeting contractual obligations, public interest requirements, safeguarding life/health, investigations by defence counsel, use of publicly available data, processing for statistical/historical purposes.

Additional provisions for legitimising transfers are laid out in Section 44 of the code and include transfers to countries deemed adequate by the European Commission, or the adoption of contractual safeguards (<http://www.garanteprivacy.it/garante/document?ID=727068>)

Guidelines for genetic biobanks have been published by the Italian Society of Human Genetics and Telethon Foundation on 2003 (<http://www.telethon.it> ; <http://sigu.univr.it>).

- Use of genetic testing and genetic information in employment and obligatory public health insurance

Data protection is regulated by the data protection law N°D.L. 196-30 giugno 2003 (CODICE IN MATERIA DI PROTEZIONE DEI DATI PERSONALI in Gazzetta Ufficiale n. 174 del 29 luglio 2003 - Supplemento Ordinario n. 123, available at <http://www.parlamento.it/parlam/leggi/deleghe/03196dl.htm>)

- Use of genetic testing and genetic information in private life and/or health insurance

Data protection is regulated by the data protection law N°D.L.196-30 giugno 2003 (CODICE IN MATERIA DI PROTEZIONE DEI DATI PERSONALI in Gazzetta Ufficiale n. 174 del 29 luglio 2003 - Supplemento Ordinario n. 123, available at <http://www.parlamento.it/parlam/leggi/deleghe/03196dl.htm>)

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

Data protection is regulated by the data protection law N°D.L.196-30 giugno 2003 (CODICE IN MATERIA DI PROTEZIONE DEI DATI PERSONALI in Gazzetta Ufficiale n. 174 del 29 luglio 2003 - Supplemento Ordinario n. 123, available at <http://www.parlamento.it/parlam/leggi/deleghe/03196dl.htm>)

Recommendations for paternity and criminal investigation testing "Raccomandazioni sulle indagini biologiche di paternita' e le indagini d'identificazione criminale" have been issued by the Italian Society of Human Genetics and the Italian Forensic Haematologists Group (Gruppo degli Ematologici Forensi Italiani-GEFI)

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

Referendum has been proposed and approved for changes in the law 40/2004, concerning medically assisted procreation. It should take place in June 2005.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

At the moment, no expert or national reference centres and/or laboratories have been identified officially on genetic testing.

In Italy there are reference centres for specific diseases (i.e. Cystic Fibrosis or Oncological reference centres), which are in charge for prevention, diagnosis (also through genetic tests), treatment and follow-up of patients.

Moreover, since 2001 there is there is a Governmental Decree (D.M. 279/2001) which establishes a National Network specific for prevention, surveillance, diagnosis and treatment of rare diseases. This network includes also some laboratories for genetic testing.

The epidemiological flow of the network is collected at the National Register of Rare Diseases, which is established in the National Centre Rare Diseases at the Istituto Superiore di Sanità.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

a) a database, including only Public Laboratories, is organized by the Istituto Superiore di Sanità in order to perform the Italian National Quality Assurance Scheme (www.cnmr.iss.it/testgenetici)

b) Italian Medical Genetics Laboratories Censes, relative to years 2002-2000-1997, have been performed and issued by the Italian Society of Human Genetics (<http://sigu.univr.it>) Censes are made on a voluntary basis of public and private laboratories which send their own data (<http://sigu.univr.it>).

7. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

In Italy, there is a Governmental Decree (D.M. 279/2001) which establishes the National Network for prevention, surveillance, diagnosis and treatment of rare diseases. This network includes also some laboratories for genetic testing.

The Italian genetic laboratories participate to the EU Network of excellence EUROGENTEST, moreover they collaborate at the OECD activities on genetic testing.

Questionnaire on genetic testing and use of genetic information

COUNTRY: LATVIA

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

The Central Medical Ethics Committee of Latvia evaluates the use of genetic testing in all research projects (requirement of the Human Genome Research Act and Latvian Council of Science). An opinion on the use of genetic testing in employment/ life and health insurance/ forensic use has not been provided.

Contact person: Dr. Laima Rudze, Secretary General of Central Medical Ethics Committee of Latvia, e-mail: laima.rudze@voava.lv or pakitis@inbox.lv.

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

A wide public debate on genetic testing and use of genetic information took place in 2001 – 2002 during the discussion on the Human Genome Research Act. The debate was organised by the group working on the project. The main conclusions were the following: 1) there is a need to develop genetic testing in Latvia; 2) interests and rights of individuals should be set higher than the interests of society and science; 3) participation of individuals in genetic testing should be completely voluntary and based on informed consent; 4) it should be prohibited to discriminate a person based on his or her genetic information.

Contact persons: prof. Elmars Grens, Director of the Biomedical Research and Study Centre (University of Latvia), e-mail: grens@biomed.lu.lv; prof. Valdis Pirags, Stradins University Hospital, Riga, Latvia, e-mail: pirags@latnet.lv.

Several conferences on ethical problems in biomedical research, incl. regulation of biobanks, data protection, and legal framework for biomedical research were also organised (main organisers: Ministry of Welfare, Central Medical Ethics Committee).

3. Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

Certification of genetic tests and genetic testing laboratories is not mandatory in Latvia. But a certificated laboratory, in accordance with the health care financing procedures specified by the Cabinet of Ministers of Latvia, has advantages when entering into contacts with medical insurance institutions. That is why most genetic testing laboratories in Latvia are certificated. Certificated laboratory have to meet certain criteria relating to quality of service, regulated by the Medical Treatment Law (1997) with amending Cabinet Regulations No. 133 (2001), 77 (2002), regulation of Ministry of Welfare No. 75 (2002). Three genetic testing laboratories regularly participate in the external quality assessment schemes run by the European Molecular Genetic Quality Network EMQN (currently in 4 schemes).

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

Use of genetic testing and genetic information in health care is regulated by the following laws: a – Medical Treatment Law (1997); b – Human Genome Research Act (2003); c – regulations of Ministry of Health on health care of pregnant women (No. 14-/13, 2004), on health care of newborns (No. 14-/13, 2004), on examination of potential germ cell donors (No. 311, 2003); d – Law of Reproductive and Sexual Health (2002).

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

Collection, storage, transmission and analysis of personal genetic information are regulated by the following laws: a – Medical Treatment Law (1997); b – Personal Data Protection Law (2000); c – Human Genome Research Act (2003).

- Use of genetic testing and genetic information in employment and obligatory public health insurance

No specific legal regulation but general prohibition in the Human Genome Research Act (2003)

- Use of genetic testing and genetic information in private life and/or health insurance

No specific legal regulation.

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

Forensic use of genetic testing is performed only in accordance with a decision taken by the investigative authority, the prosecutor or the court (judge) in accordance with the procedures prescribed by law (Civil Law, art. 245 – 250, Criminal Law)

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

Regulation of the Ministry of Health on genetic testing (based on the Human Genome Research Act).

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

No national reference centres, no national expert laboratories.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

No database on laboratories performing genetic tests.

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

No national network on genetic testing of rare genetic diseases.

Questionnaire on genetic testing and use of genetic information

COUNTRY: LITHUANIA

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

The Act No. VIII-1679/2000 of the Parliament of the Republic of Lithuania has passed the Law on Ethics of Biomedical Research, which regulates genetic testing in humans. Genetic testing in Lithuania can only be carried out for medical, scientific and forensic purposes. Research projects dealing with genetic testing of individuals from Lithuania must be approved by the Lithuanian Bioethics Committee. The web address of the Lithuanian Bioethics Committee is: <http://bioetika.sam.lt>

The Parliament of the Republic of Lithuania has ratified 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 (Oviedo, 04.IV.1997).

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

3. **Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate contact person(s).**

No specific legislation for genetic testing exists. The use of genetic testing in health care is regulated in the same way as other medical testing by the Law on Health System of Republic of Lithuania and related documents. Research projects involving genetic testing of individuals must be approved by the Lithuanian National Committee on Biomedical Ethics.

The storage and treatment of personal data is regulated by the Law on Legal Protection of Personal Data of Republic of Lithuania, adopted on 21 January 2003, No. IX-1296.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

No specific legislation regarding genetic testing exists. All laboratories offering diagnostic testing has to be certified by Service of National Accreditation for Medical institutions of Ministry of Health of Republic of Lithuania. The internal quality control scheme is required for all laboratories.

External quality assessment for genetic testing is voluntary.

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

No specific legislation for genetic testing exists. The use of genetic testing in health care is regulated in the same way as other medical testing by the Law on Health System and related documents.

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

No specific legislation regarding personal genetic information exists. The storage and treatment of personal data is regulated by the Law on Legal Protection of Personal Data of Republic of Lithuania, adopted on 21 January 2003, No. IX-1296.

- Use of genetic testing and genetic information in employment.

- Use of genetic testing and genetic information in obligatory public health insurance and private life and/or health insurance

According to the Law on Insurance of the Republic of Lithuania, adopted on 18 September 2003 No. IX-1737, the insurer is prohibited from requesting in any form the policyholder, insured person and other persons to provide to him data of genetic testing.

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

No

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

There are no national reference centres and/or national expert laboratories on genetic testing in Lithuania.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

No such database exists.

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Networking on genetic testing of rare diseases does not exist in Lithuania

The Dept. of Human and Medical Genetics of Vilnius University and the Center for Medical Genetics of Vilnius University Hospital Santariskiu Clinics participate in following EU networks/programmes: CRMGEN, ORPHANET.

Questionnaire on genetic testing and use of genetic information

COUNTRY: NETHERLANDS

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

The Dutch Health Council, a scientific advisory body to the Minister of Health has dealt with the issues in its report Heredity: Science and Society, 1989. Whenever relevant subsequent reports on specific issues have dealt with the use of genetic testing and information outside health care. The Dutch policy in this field is mainly based on the 1989 report. You will find further relevant information in the report “the application of genetics in the health care sector” the Minister of health prepared for parliament (2001). In this reply to your questionnaire I refer to that report

Following that report, on the request of the Minister of health, ZonMW (an organisation addressing issues from the perspective of policy, health research and practice) organised a so-called “consultation” with sectors involved and issued on that basis a report “application of genetics in health care” in 2003. The report addresses the implications developments in genetics may have for legislation. The issues mentioned in this question are all dealt with. It is available in Dutch only from ZON/MW e-mail: er@zonmw.nl web: www.zonmw.nl

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

See page 67 and 68 of the 2001 report on the application of genetics in the health care sector. There have been no new developments since except for the creation of the Forum for biotechnology (also mentioned in the report) that is supposed to be the liaison between society, health field and policymakers. It issues opinions and gives advice on a variety of subjects, including specific aspects related to genetics.

3. **Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate contact person(s).**

a. *The Dutch legislative policy does not focus on developing legislation specific for genetics. Genetic testing and genetic information are included in the relevant general health laws (on patient rights, quality of health services, health care professionals, insurance, medical devices etc etc). Genetic related aspects are only specifically addressed in so far as the general framework does not suffice.*

For instance the Dutch data protection act includes medical data which are considered sensitive data, and specifies that in principle personal genetic information may not be processed for other purposes than for the person concerned (except in case of an overriding health interest of another person or necessary for scientific research where the consent rule applies).

- b. *The Medical Examinations Act limits the possibility for employers and private insurance companies (except health insurance) to require inter alia a genetic test and to ask for existing (family) information in case of serious disorders/diseases without a cure available. The law is based on both protection of private life as well as on preventing barriers for access to health care. The provisions include a prohibition of medical examination for the purpose of detecting/predicting the possibility of future development of a serious disease or disorder for which no cure is available; asking for such information, if it already exists (including family and except in case the illness is manifest with the candidate insurance taker) is only allowed for when the insurance asked for exceeds a certain sum. (see p. 32-33 of the 2001 report).*

For employment the general rule is that medical examinations focused on future health risks except expected invalidity within 6 month, are not allowed prior to employment, but only after the decision has been taken. The medical examination then must focus on specific health related risks of the job, employers are expected to take preventive measures and if needed to offer another job.

The nature and extent of the applicability of the equal treatment act (=non-discrimination) to genetic discrimination is as yet not very clear.

Recently, the Dutch health Council has been asked to give advice on the implications/complications which exist particularly, but not exclusively, when the introduction of a screening programme is envisaged because from a medical perspective, a disease is considered treatable (but not per se leading to cure) while private insurance companies have a different perspective regarding the notion of "treatable". In that case each time specific arrangements have to be made with insurance companies to reach acceptable solutions for persons who somehow depend on private life insurance etc. (as has been done for instance with Hereditary FH).

With the prospects of a new system for health insurance (less public, more private) starting in 2006, the problem is likely to become more serious.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

On quality assurance please refer to chapter V, pages 47-49 and p 60 of the 2001 report. Moreover the Act on quality in health care setting rules for good quality delivery of health care in general also applies to genetic services.

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

In relation to genetic testing and the use of genetic information within health care, the Medical Treatment Act (on rights of the patient in general) is relevant, in particular as

regards information, consent and the patients right to medical secrecy. (see p. 3542 of the 2001 report).

The population screening act requires a license for screening that involves a serious disease of disorder for which no treatment or prevention is available. The act is presently under review. See also p. 53 of the 2001 report.

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and biobanks

Legislation on the use of socalled left over bodily material for research purposes is under preparation. It will also include the donation of material for biobanks. Such legislation will complement the act on medical research on human beings.

- Use of genetic testing and genetic information in employment.
(see under part b)

- Use of genetic testing and genetic information in obligatory public health insurance and private life and/or health insurance

The present public health insurance system is such that genetic testing and genetic information is not an issue. However, the more the system becomes privatised it is expected to become an issue(see also under part b)

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

Forensic use of DNA has been included in the penal code; there is a special forensic DNA bank.

h. Legislation on the use of socalled left over bodily material for research purposes is under preparation. It will also include the donation of material for biobanks. Such legislation will complement the act on medical research on human beings.

SEE FURTHER DETAILS IN ANNEX “THE APPLICATION OF GENETICS IN HEALTHCARE SECTOR CONTENTS” PREPARED BY THE MINISTRY OF HEALTH, WELFARE AND SPORT

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Questionnaire on genetic testing and use of genetic information

COUNTRY: PORTUGAL

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

- *Report and Statement on “personal genetic information” (37/CNECV/01)*
- *Report and Statement on “the ethical implications of genomics” (40/CNECV/01)*
- *Report and Statement on “personal genetic information” (43/CNECV/04)*

Contact: *Dr. Paula Martinho da Silva,
President,
Concelho Nacional de Ética para as Ciências da Vida (CNECV)
Email: pres.cnecv@sg.pcm.gov.pt
<http://www.cnecv.gov.pt/CNECV/>*

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

There have been several public debates, over time, organized by scientific and medical institutions, scientific and professional societies, the national medical association and the parliament

3. **Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.**

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

None specific to genetic testing

IPQ (Portuguese Institute for Quality) is the general official body for accreditation

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

Law 12/2005 (26 January) regulates the use, storage, property and circulation of genetic information and of biological samples, both for testing and research purposes

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

All covered under Law 12/2005, 26 January 2005 (see Appendix)

- Use of genetic testing and genetic information in employment and obligatory public health insurance

Covered under Law 12/2005, 26 January 2005

- Use of genetic testing and genetic information in private life and/or health insurance
Covered under Law 12/2005, 26 January 2005

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

No legislation adopted so far

In addition, the following regulation from the Ministry of Health exists:

Prenatal Diagnosis – Despacho 5411/97 (2ª série), DR n° 180, 6/8/97; and Despacho n° 10325/99 (2ª série), DR (II série) n° 122, 26/5/99

Genetic Testing – Despacho 9108/97 (2ª série), DR (II série) n° 237, 13/10/97; and Portaria 189/98, DR (I série B) n° 68, 21/3/97

*Contact: Prof. Guilherme de Oliveira/Prof. André Pereira
Centro de Direito Biomédico
Faculdade de Direito, Univ. Coimbra
Emails: goliv@fd.uc.pt and andreper@fd.uc.pt*

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

Yes, specific for preimplantation genetic diagnosis (PGD)

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

There is a Network for Hospital Reference in Medical Genetics, recently created by the Ministry of Health (Direcção-Geral de Saúde). It includes only institutions of the national health system, leaving out private, university and research labs and institutions.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

- *Portuguese Society for Human Genetics (molecular genetics club)*
- *College of Medical Genetics, at the Portuguese Medical Association*
- *Direcção-Geral de Saúde (contact person: Dr. Beatriz Calado)*

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Only in an informal manner
A national chapter of Orphanet also exists

Law 12/2005 (26 February)

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

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

Defines medical information as the health information used for medical intervention

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

The same holds true also for predictive/pre-symptomatic testing. Pharmacogenetic testing is not provided at present. Nation wide newborn screening is provided for phenylketonuria and hypothyreosis.

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

This topic will be addressed in the prepared new legal framework.

- Use of genetic testing and genetic information in employment.

This topic will also be addressed in the prepared new legal framework.

- Use of genetic testing and genetic information in obligatory public health insurance and private life and/or health insurance

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

This topic is regulated by law No. 417/2002

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

A new legal framework is under preparation.

5. Could you please indicate if national reference centers and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

At present there are no national reference centers and/or national expert laboratories on genetic testing in Slovakia.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

No such database exists. But there is an established network of departments of medical genetics, which are in contact with the few laboratories providing genetic testing.

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

No such networking exists, DNA testing for rare diseases is provided on the basis of personal contacts.

Questionnaire on genetic testing and use of genetic information

COUNTRY: SPAIN

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

Not in a formal way, but there have been many discussions in scientific meetings and several newspapers have published information on the importance of molecular genetic tests in the diagnostic of some diseases and in forensic identification of corpses, among other related aspects.

3. **Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.**

Legal framework for:

There is no specific legislation on genetic testing. Other legislation is applied such as the Human Rights and Biomedicine Convention Agreement; in force since 1st January 2000 ruling the use of genetic information for medical or medical research purposes and the Law of Personal Data Protection

-Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

There are only unofficial initiatives

- Use of genetic testing and genetic information in employment and obligatory public health insurance

The Spanish Constitution (Art. 43) provides the right to public health protection to all people.

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

Forensic use of genetic testing is ruled by the Spanish Law 15/2003 (25th Nov. 2003) in Art.s 326 and 363 with the specific requisites any genetic testing has to accomplish to be acceptable by the Courts of Justice.

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

A new law is at the moment being prepared by the Spanish Ministry of Health that will include specific rules for genetic testing and the use of genetic information.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

There are regional and national “reference laboratories” on genetic testing, especially for rare diseases. However due to the lack of specific legislation on the topic they are not officially appointed as such they are only “de facto”.

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

There is no at the moment, a database on Spanish laboratories performing genetic tests, but it has been recently published a “Directory of Spanish Molecular Testing Laboratories” that includes detailed information of 92 laboratories.

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

There are at least two networks at national level on genetic testing on rare diseases:

a) Network INERGEN, Coordinated by Dr. Gillermo Antiñolo (Sevilla)

b) Network RECGEN, Coordinated by Dr. Luis Perez Jurado (Barcelona)

Both networks are included in the European Union Network EUROGENTEST financed by the CEE 6FP and Coordinated by Prof. J. J. Cassiman.

Questionnaire on genetic testing and use of genetic information

COUNTRY: SWEDEN

A Commission on Genetic Integrity was set up. They delivered their report a year ago. Their proposals will result in a Bill for Parliament. For their proposals see attached summary.

1. Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?

X yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

Swedish National Council on Medical Ethics

<http://www.smer.gov.se/>

2. Will (or has) a public debate take (n) place on genetic testing and use of genetic information?

XX yes, some no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?
Workshops, hearings with lay people etc. Details and reports are found at The Swedish gene technology Advisory Board

<http://www.genteknik.se>

homepage.

*Contact person: Gustaf Brunius **email** genteknik@genteknik.se*

3. Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate a contact person.

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

No specific regulations

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

Law on genetic screening using DNA (see attached text)

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

Law on biobanking

Law on bioethics in research

Law on registration of personal data

National death registry

Swedish cancer registry

National malformation registry

- Use of genetic testing and genetic information in employment and obligatory public health insurance

No regulations, new legal regulations under preparation

- Use of genetic testing and genetic information in private life and/or health insurance

Agreement between the national insurance companies and the government, however the Commission on Genetic Integrity has proposed legal regulation (see attached summary)

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

Regulation exists. New legal regulations under preparation

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

SOU 2004:20, State public committee on Genetics, Integrity and Ethics. A published document as a basis for future legislation. Summary in English.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

Not existing

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

On the website of the Swedish Society for Medical Genetics

<http://www.svls.se/sektioner/mg/index.htm>

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Yes, there is a tight connection between the six clinical genetics centres.

Participation in European EQA schemes are available

Listed in EDDNAL and Genetests

FURTHER INFORMATION IN ANNEX

Questionnaire on genetic testing and use of genetic information

COUNTRY: UK

1. **Has your National Ethics Committee or similar body provided an opinion on the use of genetic testing/ genetic information in research/health care/employment/ life and health insurance / forensic use (e.g. criminal investigation and public security)?**

yes no

If yes, could you please summarise the recommendations, indicate the contact person and web address from where the extended version of the opinion(s) is/ are available:

The Human Genetics Commission is the UK's Government's Advisory Body on developments in human genetics and how they impact on individual lives. The recommendations (attached at Annex A) are taken from the HGC's report 'Inside Information', which can be viewed at: <http://www.hgc.gov.uk>

The UK operates a distributed system of ethical review, and a great number of other national organisations – Parliamentary and non-governmental – have published reports on issues relating to the above uses. Notable examples include the Nuffield Council on Bioethics report in 1993 on genetic screening and the 1995 report on human genetics by the House of Commons Select Committee on Science and Technology. The latter report led to the setting up of the Human Genetics Advisory Commission and the Advisory Committee on Genetic Testing which merged to form the Human Genetics Commission.

2. **Will (or has) a public debate take (n) place on genetic testing and use of genetic information?**

yes no

If yes, how was / will it be organised, what were the conclusions, who is the contact person?

In the UK these issues have been debated in many fora over many years, and this will no doubt continue. The UK government is committed to ensuring greater public understanding of genetics through transparency and openness and encouraging wider public debate.

For 'Inside Information', members of the public were invited to comment on the issues raised in a public consultation, which formed the recommendations that were made in the document. The consultation was preceded by a survey of public attitudes to genetic information. HGC also has a Consultative Panel of people affected by a genetic disorder which comments on all aspects of the Commission's work.

3. Could you please describe the current legal and / or regulatory framework for genetic testing and use of genetic information in your country and indicate contact person(s).

Legal framework for:

- Quality assurance of genetic tests and testing services (including license/accreditation requirements specific for genetic testing and official quality assessment systems in place)

There is no formal legal framework for laboratories in the healthcare system, but NHS laboratories are required to have or be seeking accreditation with Clinical Pathology Accreditation (UK) Ltd (CPA) (or another relevant body accrediting to equivalent standards. CPA provides a means to accredit Clinical Pathology Services and External Quality Assessment Schemes (EQA). It involves an external audit of the ability to provide a service of high quality by declaring a defined standard of practice, which is confirmed by peer review.

Most NHS laboratories providing molecular genetic tests are members of the UK Genetic Testing Network (UK GTN) which is a network of laboratories that offer molecular tests for inherited single gene germ line disorders. All UKGTN laboratories adhere to quality criteria and standards, including CPA or equivalent accreditation, and work within clinical governance arrangements. In addition the tests offered on the network undergo a process of evaluation to ensure scientific validity and clinical utility. The robust entry requirements and test evaluation procedures ensure that UKGTN laboratories offer an efficient and effective quality service that meets the needs of both patients and clinicians.

- Use of genetic testing and genetic information in health care (including for diagnosis, screening of newborns and adults, predictive/ pre-symptomatic testing, pharmacogenetics, selection of donors ...)

Genetic testing in clinical care is covered by a combination of professional codes and guidance on standards for counselling, consent and appropriateness of testing. This is implemented locally via clinical governance within hospitals and trusts. There are a variety of sanctions for breaking these standards, including disciplinary procedures by the General Medical Council or court action under common law duties for consent, confidentiality or breach of duty of care.

From the 2006 the Human Tissue Act 2004 will require consent before the storage or use of tissues for healthcare or research. It includes a specific criminal offence of testing a person's DNA without consent (so-called "DNA theft").

The main provision relating to clinical uses of genetic information is the Data Protection Act 1998 that implements the corresponding Directive.

- Collection, storage transmission and analysis of personal genetic information for the purpose of public health and/or medical research including applications like medical registers and bio-banks

In England and Wales, the Human Tissue Act includes provisions relating to the storage and use of human tissue for the purposes of scientific research. It requires specific consent to use tissue for research, unless the tissue is anonymised and used for research in a manner approved by a Research Ethics Committee. Similar legislation is under consideration in Scotland and Northern Ireland.

Acts and regulations implementing EU Directives on matters such as data protection or clinical trials will also regulate the use of personal genetic information in research contexts.

All research undertaken in National Health Service premises, involving NHS employees, facilities or patients, tissue or data collected from NHS patients must comply with a stringent Research Governance framework that sets out the statutory duties, professional obligations and the requirements for ethical research.

- Use of genetic testing and genetic information in employment.

The Data Protection Act 1998 is supported by statutory Codes of Practice and guidance, including specific guidance on employer's use of employee's genetic data. This incorporates guidance from the HGC. It advises, under the principles of fair processing, that employers:

- *do not use genetic testing as a predictor of future ill health or possible time off work;*
- *do not insist that a worker discloses the results of previous genetic tests and*
- *only use genetic testing to obtain information when it is clear that a worker with a predictable genetic condition is likely to pose a serious safety risk to others or the working environment may place them at a specific risk.*

- Use of genetic testing and genetic information in obligatory public health insurance and private life and/or health insurance

There is a moratorium on the use of genetic tests by insurance companies. The 5-year genetics and insurance Moratorium was agreed with the ABI in November 2001. Under the moratorium, insurance companies can only ask for the results of predictive genetic tests if (a) the test has been approved by the Genetics and Insurance Committee (GAIC) and (b) the policy is for more than the financial limits of £500k of life cover or £300k for other types of health insurance (critical illness, income protection). So far GAIC has only approved the use of Huntington's disease genetic test results in setting life insurance premiums.

The moratorium has now been in force for 3 years and the Government is taking into account the views of the ABI, GAIC and HGC on a longer term solution.

- Forensic use of genetic testing in criminal investigation and public security (including fights against terrorism)

The taking, use and retention of DNA samples from suspects in the investigation and prosecution of crime is covered by the Police and Criminal Evidence Act 1984, as amended. The National DNA Database holds approximately 2.8 million DNA profiles taken from

persons who have been arrested for, charged with, informed they will be reported for, or convicted of a recordable offence. The database also holds approximately 245,000 profiles obtained from crime stains left at scenes of crime.

The National DNA Database is a vital police intelligence tool helping to identify offenders more quickly, make earlier arrests and secure more convictions. DNA evidence has transformed the fight against crime, helping to catch both serious offenders and those committing "volume" crime, like burglary or car theft, where it's impact has been greatest. In domestic burglary the detection rate rises from 14% overall to 45% where crime scene DNA is put on the National DNA Database. Serious offenders eg sex offenders and other violent offenders are often detected because they are arrested and sampled at a later date for a relatively minor offence. One example is a case where in 1998, a rape and indecent assault were carried out on an 11 year old and her 9 year old sister. In 2001, a shoplifter was arrested in another town many miles away and a DNA sample was taken. The shoplifter's DNA was found to match the 1998 crime scene profiles. The offender pleaded guilty to the offences and was sentenced to 15 years imprisonment.

Advances in scientific techniques also mean that the evidence in previously unsolved crimes can be re-examined and offenders detected many years after the crime was committed.

4. Could you please indicate if any new legal and/or regulatory framework for genetic testing and use of genetic information is under preparation/discussion?

No, but the genetics White Paper ("Our inheritance, our future - realising the potential of genetics in the NHS", which was launched in June 2003, <http://www.dh.gov.uk>

- set out the safeguards that the UK government has already introduced (eg the moratorium on unacceptable use of genetic tests by insurers, banning human reproductive cloning and regulating the use of genetics in reproductive medicine and clinical trials of gene therapy)*
- recognised that developments in genetics would continue to present new ethical challenges*
- committed to ensuring that the regulatory framework anticipates and continues to address public concerns.*

Since then the UK government has successfully passed the Human Tissue Act 2004 which makes it a criminal offence to obtain bodily material with the intention of testing a person's DNA without proper consent. This offence applies across the UK.

5. Could you please indicate if national reference centres and/or national expert laboratories on genetic testing exist in your country and which criteria have been used to select these?

There are 2 national genetics reference laboratories (NGRLs) Manchester and Wessex (based in Salisbury) in England. The national reference function was added to these two well-established laboratories in 2002.

Their remit includes:

- *Anticipating the development of new genetic technologies and carrying out Health Technology Assessment*
- *Developing new and better ways of testing, including realising the diagnostic potential of gene discoveries when no other network laboratory is involved*
- *Taking a lead in developing quality assurance systems by working with independent QA agencies, developing internal quality control procedures and collaborating in the development of new reference materials for genetic testing*
- *Providing diagnostic services for families with extremely rare disorders where the mutation has already been characterised and no other network laboratory can offer the service*
- *Identifying better ways of working in laboratories e.g. skill mix, use of robotics, and laboratory organisation. The reference laboratory will be expected to develop, and provide a resource for, best practice for service provision and will be involved in developing innovative working practices to improve reporting times and cost-effectiveness.*
- *Education and training for staff in genetics and other specialties – to ensure dissemination of knowledge and capacity in the new methodologies and technologies*
- *Providing support for the national genetic testing network and working with professional bodies on disseminating information on the network and in further developing audit across the network*

The two laboratories were selected as a result of a competitive process. Criteria for selection included:

- *Solid institutional base – established institutions, with strong university and NHS links and structures for developing and recruiting high quality staff, and a record in attracting funding from recognised sources*
- *Multi-disciplinary scientific skills*
- *R&D management experience, plus administrative capabilities*
- *Appropriateness of the budget to achieve the stated objectives, access to effective financial and accounting systems and the availability of other funding streams and their use to develop programmes of added value.*
- *The overall business plan*
- *Facilities available.*

6. Could you please indicate if a database on laboratories performing genetic tests exists in your country?

The UK GTN has produced an inventory which lists diseases for which molecular genetic tests are available under the NHS and the UKGTN laboratories that offer testing for them. It has been compiled from information collected in May 2003 and is an interim measure prior to a searchable database being available on the UK GTN website (www.geneticstestingnetwork.org.uk/). Once the database is available it will allow users to

find what UKGTN laboratories are providing what tests. It will be searchable by disease, gene, by OMIM number or laboratory and will be regularly updated so the information is current.

8. Could you please indicate if networking on genetic testing of rare diseases exists in your country and if these networks collaborate at EU / international level?

Networking between labs and 'exporting' and 'importing' of tests between UK labs is well-established practice. This has become more formalised since the UK GTN was set up. One of the purposes of the UK GTN is to support rational provision of testing and the movement of rare tests around the network and to ensure tests are available at the optimum number of labs to allow efficient use of resources and to provide a consistent and reliable service.

UK labs also have long-established links internationally (both with Europe and globally). This is to support import and export of extremely rare tests but also to share scientific and clinical knowledge. UK participates in Eurogentest and many other networks, some of which are disease-specific eg the European Skeletal Dysplasia Network.

Relevant web sites:

UK Genetic testing Network:

www.ukgtn.org

The national genetics reference laboratories:

<http://www.ngrl.co.uk/Pages/index.htm>

and the public health genetics unit:

<http://www.phgu.org.uk/index.php>

Genetic testing in the UK

In common with many Member States, the UK has recognised the importance of development in genetics in the past decade or so. The clinical benefits and potential ethical problems raised by predictive genetic tests have been extensively debated.

The UK Government and Parliament have conducted several significant reviews and reports since the middle of the 1990's – details are given elsewhere. There are two recent examples, the first is a report by the Human Genetics Commission on the ethical, legal and social aspects of genetic information, published in May 2002. It coincided with the announcement of a extra investment in genetics in the National Health Service in England and for genetics research, as well as legislation and policy initiatives to protect against misuse of genetic information. A major policy statement – the Genetics White Paper "*Our inheritance, our future - realising the potential of genetics in the NHS*", was published by the Department of Health in June 2003.

This announced a £50 million programme of investment to help prepare the National Health Service to make appropriate use of genetic knowledge and technologies as they emerge. Over £20 million of this has already been allocated to modernisation of genetic laboratories in England.

Genetics in healthcare is devolved to the Scottish Parliament, the National Assembly for Wales and to the Northern Ireland Assembly. They are conducting similar reviews and extra investment to improve on the already good networking between the four countries in the UK.

Organisation of services

There are **around 25 specialised genetics centres** in the UK. Most laboratories are linked to a clinical service.

The vast majority of NHS genetic testing is provided from service labs but a tiny proportion of tests (eg very rare conditions) are done in research labs (often the mutation is then confirmed in a service lab). UK laboratories send a small number of samples abroad for testing and also test samples from other countries.

The UK Genetic Testing Network is an informal network of most molecular service labs in UK. It was set up in 2002 with a multi-professional steering group and is run by a team of staff. It builds on long-standing collaboration between centres. Its main aim is to promote equity of access to tests. Laboratories have to satisfy quality criteria. UKGTN assesses tests according to criteria of scientific validity and clinical utility before recommending them for funding on the NHS. The UKGTN website contains a list of approved laboratories and a list of approved tests.

Two **National Genetic Reference Laboratories** (in Manchester and Wessex) were established in 2002. These were selected to undertake a special work programme (research, training and technology assessment). They were *not* selected to be “the best” lab for any given test or disease.

UK laboratories and clinicians have **collaborated internationally** for many years. Some UK laboratories are members of the European Molecular Genetics Quality Network (EMQN) and CRMGEN (Certified Reference Materials for Molecular Genetic Testing). But collaboration goes beyond the EU and involves experts in eg Australia, US, Canada.

Population screening programmes are being implemented for Downs syndrome, sickle cell, thalassaemia and cystic fibrosis. New ideas for screening programmes have to satisfy the criteria of the National Screening Committee, and these include some new criteria for genetic testing.

Quality

In the UK there are 6 levers all acting now to increase quality:

- *All* pathology labs are strongly encouraged to achieve accreditation (through Clinical Pathology Accreditation Ltd or equivalent)
- For genetics laboratories, there have been two rounds of capital investment from government (in 2001 and 2004) to improve quality and capacity
- Government funding has been channelled through local service funders who have taken more interest in quality and capacity of their services as a result
- Laboratories must be accredited to be a member of UKGTN

- Professional bodies have active discussions and guidelines on best practice
- Rationalisation of testing between labs allows concentration of expertise and better economies of scale

Use of **external reference standards** is required for CPA accreditation. But there is a need for a wider range of these. In addition to CRMGEN, development of new external reference standards is part of the National Genetics Reference Laboratory work programme.

Legislation and protection

Following recommendations from Parliament and the Human Genetics Commission, the Government has responded to concerns about the potential for misuse of genetic information and has, where possible, adopted effective non-statutory approaches that can be adapted to the fast moving developments.

One exception is the testing of DNA without proper consent. The Government has recently passed legislation making it a criminal offence to obtain bodily material with the intention of analysing DNA without consent. This is intended as a deterrent in intrusive cases of disputed parentage or for other unacceptable purposes. It does not affect legitimate uses for clinical care, research or forensic uses.

In 2001 the Government and the UK Association of British Insurers have agreed a five year moratorium on the use of predictive genetic test results for life and health insurance. This is currently being reviewed.

The Office of the Information Commissioner has also recently issued a statutory code of practice on the use of genetic information by employers which makes it clear that in most circumstances it is unacceptable to demand or offer genetic testing or to require disclosure of test results taken for clinical purposes.

In the UK developments in genetics are kept under review by bodies such as the Human Genetics Commission who encourage wide public dialogue. The UK is also active in discussions at international forums such as the European Commission, the Council of Europe, the United Nations, for example UNESCO. We are particularly pleased to support the important work being conducted by the OECD following the Vienna conference in February 2000. This has already improved our understanding on important matters such as the organisation and volume of genetic testing in the EU and other developed countries.

Dianne Kennard
Mark Bale
Department of Health, London

Table of Recommendations in HGC Report May 2002

2.07 (2.7)	One such instrument [to establish a common international framework] which we believe to be particularly important is the Council of Europe's Convention on Human Rights and Biomedicine, and we recommend that the Government take steps towards signing and ratifying this Convention.
2.11 2.20	Genetic knowledge may bring people into a special moral relationship with one another. We have therefore proposed the following concept of genetic solidarity and altruism, which promotes the common good. We share the assumption of our society that individual persons have the highest moral importance or value. This requires us to be sensitive to the special role that genetic identity has come to play in people's lives. We [outline] the key principle of respect for persons.
2.22	We believe that a number of secondary principles may be derived from this overarching principle, taking account of the possible claims of genetic solidarity and altruism: the principles of privacy, consent, confidentiality, non-discrimination.
3.24	We recommend that best practice for clinicians is that, however remote, all possibilities [relating to the outcome of a genetic test], should be discussed with both individuals before consenting to the test.
3.30	However, we have taken note of comments in response to our consultation and wish to give further consideration to pharmacogenetic testing, especially to the wider social and ethical implications of its increased use in regulation and prescribing of medicines.
3.36	We do not believe that it is feasible for separate arrangements to be made for the storage of genetic information within the health service, but nonetheless we point out that the potentially sensitive nature of this information underlines the importance of protecting the confidentiality of patient medical information in general.
3.37	We intend to monitor any future schemes for the ready storage and accessing of genetic information for prescribing purposes.
3.39	We believe that the requirements of medical confidentiality need to be clearly understood, at all levels and across the entire medical and biomedical research field. Maintaining confidentiality should become an essential part of employment contracts and of membership of relevant professional bodies. This should be backed by sanctions.
3.48	We comment on the possible need for a broader offence against breach of medical confidence, which is beyond the scope of this report.
3.60	We recommend that consideration be given to the creation of a criminal offence of the non-consensual or deceitful obtaining and/or analysis of personal genetic information for non-medical purposes.
3.61	We believe that it would be sensible to conduct our review of direct access to genetic tests in the light of our recommendation relating to a new criminal offence.
3.62	The general duty to maintain the confidential nature of personal genetic information is not an absolute one. We note circumstances where it may not be appropriate, such as where consent is given or where it is in the interest of the patient, of relatives, or of the wider public
3.68	Bearing in mind the principle of genetic solidarity and altruism, we take the view that the disclosure of sensitive personal genetic information for the benefit of family members in certain circumstances may occasionally be justified. This would arise where a patient refuses to consent to such disclosure and the benefit of disclosure substantially outweighs the patient's claim to confidentiality
3.72	We believe that in exceptional cases it should be permissible to reveal personal genetic information in order to avert substantial harm to others

4.01 (4.1)	Although we understand the concerns of relatives over the revealing of family medical history, we believe that it is proper for questions in this area to be asked by clinicians and genetic counsellors, and we believe that the patient has a right to disclose it
4.07	We believe that there may be a need for secondary legislation to ensure that the holders of information about genetic relatives in a clinical context are specifically exempted from their normal obligations of notification and provision of information to such relatives under the Data Protection Act.
4.15	Carrier testing is used to determine whether a person carries the gene for a recessive genetic condition. Consent to this form of testing requires information about the implications of carrier status, both in terms of the psychological and social impact on the affected person and in terms of the implications it may have for reproductive decisions
4.21	We intend to consider the matter of prenatal genetic testing in more detail in our work on genetic testing and reproductive choice.
4.26.1 (4.26)	At present, we recommend that where multiple genetic tests are carried out, it should be explained to the patient what the principal purpose of testing is, and that it will reveal additional information.
4.26.2 (4.26)	We recommend that before new multiple genetic tests are introduced, the implications for adequately informed consent are considered carefully.
4.32	The placing of a large amount of genetic information about a person on a single electronic record could also pose a challenge to privacy. We conclude that there are no persuasive reasons to resist the introduction of new information storage technologies even if these do present some challenges to confidentiality. At the same time, we stress the importance of incorporating the traditional principle of medical confidentiality into the regulation of such technologies
4.35	Genetic screening programmes are important in preventing ill health, but they raise certain ethical issues which we discuss in this report. We shall be monitoring developments in national screening programmes and will be considering screening issues as part of our planned work on genetics and reproductive choice
4.38	There are particular legal and ethical issues involved in those cases where consent cannot be obtained from the person being tested. We endorse the recommendation of the Advisory Committee on Genetic Testing that great caution should be observed in the testing of children for late-onset disorders
4.57	We conclude that benefit to a relative, and hence indirect benefit to the interests of the tested person, should be factors to be taken into account in deciding whether genetic testing should be carried out on a person who is unable to consent to it
4.67	There may be some clinical situations where genetic information about the dead is needed in order to assess a risk to a living relative. This information may be obtained by testing samples removed from an individual during life. The approach we favour is that a presumption should be made that the dead person would have consented in his or her lifetime to such testing and that this justifies post-mortem testing
4.73	In other cases, if testing of samples from the dead is not justified by weighty reasons such as the significant interests of other family members or of the wider public, then such testing should be regarded as unethical
5.1	Our understanding of genes and of how they work in the human body is the result of prolonged and extensive research efforts. If this understanding is to be translated into therapeutic benefit, such research must be given every encouragement. Genetics is a vital part of this and we therefore all have an interest in successful genetics-based medical or health-related research.
5.13	We recommend that the Government gives a firm commitment to funding research and development initiatives on this important aspect [satisfactory techniques of encryption] of data security

5.14	The very nature of DNA limits the process of complete anonymisation, because it may be possible to link a sample by use of “DNA fingerprinting”. We nonetheless feel that for practical purposes the concept of anonymisation is valid.
5.15	The need to obtain the consent of the participant at the outset is a fundamental principle of ethical research. We devote considerable attention to issues that must be addressed in consent procedures for different types of research.
5.19	We acknowledge the importance of initial consent, but consider that repeated processes of re-consent for subsequent use are impractical and, moreover, may be unnecessarily intrusive. We therefore consider that it is acceptable to seek general consent where there is to be anonymisation of data and samples. We consider that specific consent may be required where data or samples are not anonymised
5.21	The Health and Social Care Act 2001 entitles the Secretary of State to authorise the use of patient information in research without seeking patient consent to this use. We note the objection that this constitutes a significant exception to the normal rule of confidentiality and to the principle that research on patient information should proceed only with the consent of the patient in question. At the same time, we appreciate the importance of such research in areas such as cancer registries. We intend therefore to monitor the use of these powers and seek to work with the Patient Information Advisory Group on this issue
5.22	In all cases, we consider that best practice requires that the consent should clearly specify the arrangements for withdrawal from the study and the subsequent fate of samples and data
5.25	We conclude that best practice requires that the question of commercial involvement in research or access to genetic databases should be fully explained at the time of obtaining participants’ consent. This should include a brief explanation of any intellectual property issues. In order to allay concern about wider uses it might be necessary to give commercial access only to companies engaged in health-related research
5.27	There are important collections of samples which were obtained in the past and which may not be covered by any donor consent to research use. It is our view that it would be undesirable to prohibit the use of such material in cases where it is not possible to trace the donors and obtain their consent. We therefore endorse the advice given by the Medical Research Council that samples from historical collections may be used subject to certain conditions
5.28	Tissue left over from surgical operations provides a rich potential source of research material. We believe that best practice requires that tissue left over from surgical procedures should only be used for research if the patient has consented. But we believe that it is acceptable to use older collections already obtained and where consent was not sought. Such samples, however, must be anonymised
5.33	We note the different ways in which ethical oversight and approval of research is provided. We do not recommend the separate regulation of genetics research, but we are aware of criticisms of the current system of ethics committee regulation, which need to be addressed. We conclude that best practice requires that all genetic research on human non-anonymised tissue samples or bodily materials should be subject to review by an independent research ethics committee and should be monitored for compliance through clearly specified arrangements
5.37	We shall be monitoring developments in national screening programmes and will be considering screening issues as part of our planned work on genetics and reproductive choice. We recommend that the Government should encourage relevant research institutions, professional bodies and funding organisations to establish clear policies aimed at ensuring compliance with emerging best practice in ethical research. We also recommend that compliance with best practice and the application to research in genetics of the new standards for the governance of research ethics committees is formally reviewed in three to five years’ time.

5.44	We think that a morally sensitive regime can and should be worked out for the use of large-scale genetic databases and we propose to continue our discussions on this, and other issues, with those responsible for BioBank UK.
5.45	We therefore recommend that the governance of genetic research databases and DNA collections should allow for oversight by an independent body – whether it is an ethics committee or another body – which is separate from the owners and users of the database.
5.49	We recommend that the operators of all genetic research databases should be required to take rigorous steps to ensure that unauthorised access or disclosures are prevented.
5.50	We recommend that genetic research databases established for health research should not be used for any purpose other than such research and that this be put beyond any doubt, by legislation if necessary.
6.1	We consider how personal genetic information obtained in a clinical or research setting is used in other areas. We have primarily looked at the use of personal genetic information in life and health insurance and in employment. Indeed, we detect close links between the two.
6.19.1	We welcome the clear statement that genetic test results obtained from research will not be used by insurance companies.
6.19.2	We would welcome assurance from the main employer groups, trades unions and professional bodies connected with occupational health and recruitment that individual genetic research results will not be considered in making employment decisions about that person
6.31	In the light of our recommendation on separate legislation to address genetic discrimination, we recommend that no further consideration be given to amending the Disability Discrimination Act to include protection for those who have a pre-symptomatic genetic condition.
6.41	There is an opportunity to consider “genetic non-discrimination” legislation as part of the review of genetic information in employment (due in 2005) and during the moratorium on the use of genetic information in insurance (to 2006). We recommend that the Government consider in detail the possible need for separate UK legislation to prevent genetic discrimination and that this evaluation form part of a long-term policy review on the use of personal genetic information in insurance and employment.
7.07 (7.7)	In May 2001 we published interim recommendations calling for a moratorium on the use of genetic information in insurance. In October 2001, the Government and the Association of British Insurers (ABI) responded to the House of Commons Science and Technology report on genetics and insurance. We welcome the Government response and action taken by the Association of British Insurers and the opportunity that this offers during the five year moratorium for a fuller discussion of the use of genetic information in insurance underwriting
7.16	We do not at present recommend that the insurance moratorium should be extended to the use of family history information.
7.17	We further recommend that in reviewing its criteria for judging applications the Genetics and Insurance Committee (GAIC) consider the evidence which the insurance industry uses to justify its use of family history evidence to set insurance premiums.
7.18	We recommend that the Government and insurance industry should continue to fund independent research on genetics and family history. We also believe that the ABI should encourage their member companies to consider publishing the results of their own research and analysis in peer-reviewed journals.
7.27	We note a possible consumer perspective that people with no adverse family history should be able to make use of their genetic information to obtain lower premiums. This ‘preferred-life’ underwriting is superficially attractive but we recommend that it

	should not be introduced into the UK insurance market and note that the ABI have recommended against this in their Code of Practice
7.28	However, we also recommend that research be commissioned to establish the extent to which patients raise insurance considerations during consultations that precede genetic tests.
7.36	In our view, the effect of [data protection] provisions is to emphasise the importance of ensuring that insurance companies request only the minimum amount of specific information about the applicant's family history that is needed to make an insurance underwriting decision
7.50	We recommend that the Government promote the mechanisms set out in the 1999 White Paper "Modern markets – confident consumers" to help establish consumer information partnerships which could provide access to affordable insurance for those with a genetic condition.
7.56	We welcome the Government's commitment that GAIC will review the criteria for judging applications to use genetic test information in insurance. We offer some comments based on responses to our consultation.
7.59	We look forward to a closer working relationship with the reformed GAIC and would welcome both formal and informal opportunities for collaboration
7.64	We recommend that during the period of the moratorium risk pooling and other models should be explored further by independent experts from the actuarial profession, the insurance industry and the genetics community.
7.65	We intend to continue to play a role in the debate and review of genetics and insurance and we shall recommend to Government a programme which will encompass this.
8.09 (8.9)	Personal genetic information may be used by an employer to make decisions about people's susceptibility to hazards in the workplace. It may also be used to determine whether employees may pose a safety risk to others or whether they may have long periods of illness or inability to work on medical grounds. Increasingly genetic information may also be used in decisions about access to employee benefits such as occupational pensions, private health insurance and other forms of insurance. Although we recognise these possibilities, we conclude at present there is no evidence in this country of any systematic use of predictive personal genetic information in employment
8.15	We generally believe that – in accordance with the principle of respect for persons – employers must not demand that an individual take a genetic test as a condition of employment
8.18	At present, we conclude that given the current uncertainties about interpreting genetic information, at present it may be more appropriate to monitor the health of a person by other, more direct, means
8.19	We do not consider that a body like GAIC should be established at this stage to decide whether particular genetic tests are relevant for employment purposes. However, we wish to give more thought to the broader ethical and social implications of this issue, with a view to advising Government in the future. We would therefore encourage a voluntary undertaking by employers or other groups to inform HGC of any proposals to use genetic testing for health and safety or recruitment purposes.
8.23	The proposed review of the use of personal genetic information in employment by 2005 is timely, but these issues should be addressed before then. We recommend that a joint Committee be formed to monitor developments in genetic testing and employment and that this committee should include representatives from HGC, the Health and Safety Commission, the Disability Rights Commission and other interested parties.
8.26	The Information Commissioner has consulted on a draft Code of Practice on the use of personal data within employer/employee relationships. We would suggest that the Commissioner clarify that the relevant parts of this code cover both the results of

	previous genetic tests and other personal genetic information (such as a family history of a genetic condition).
8.27	We believe that any future discussions on the use of genetic information in private insurance should also consider the wider implications for employment-benefit schemes that are based wholly or partly on financial products that are based on the recognised principles of private insurance underwriting.
9.15	We recommend that consideration should be given to the adequacy of arrangements to ensure that Scottish DNA profiles and samples on the UK National DNA Database are handled in accordance with the provisions of the relevant Scottish legislation.
9.20	We would urge the Government to promote a greater degree of public dialogue about the justification for the apparent increase in the range of offences for which samples may be taken.
9.26	We recommend that the Home Office guidance and police consent forms clearly set out the important differences between the two consents that are being obtained. Where possible the obtaining of consent to provide the initial elimination sample should be separated physically and/or temporally from consent to retain the sample and profile.
9.32	We recommend that, at the very least, the Home Office and Association of Chief Police Officers establish an independent body, which would include lay membership, to oversee the work of the National DNA Database custodian and the profile suppliers.
9.39	We also draw a clear distinction between the retention of the DNA profile (the “DNA fingerprint”) on a computer and the original sample (the “CJ sample”) in a freezer. The latter may potentially be retested and used in ways not considered when it was originally taken. We welcome the Government’s willingness to consider an independent oversight body for CJ samples. We recommend that any review leading to the establishment of such a body should have a sufficiently broad remit to consider first whether or not CJ samples should be retained.
9.45	We note the important research into the use of DNA techniques in crime detection. This includes ways of identifying commonplace characteristics so that in the future a “genetic photo-fit” could be built up from a sample left at the scene of a crime. We recommend that in the short-term the Home Office and Forensic Science Service establish an independent research ethics committee to approve such research.
9.48	It appears to us that there is a clear distinction between using DNA for comparison or identification purposes (which the public broadly accepts) and using it to predict the characteristics of a person. We take the view that the public might have concerns about such uses. We therefore recommend to the Government that any proposal to use sensitive personal genetic information for forensic purposes should be subject to a full public debate in order to examine the ethical, consent and confidentiality issues.
9.55	We share general concerns about police access to research genetic databases, both from an ethical and a scientific point of view (as this may affect the range of volunteers who are prepared to participate in such research). We therefore recommend that consideration be given to legal means of preventing access to biomedical genetic databases by police and other law enforcement agencies.
10.20	A large amount of paternity testing is done for Government purposes, by bodies such as the Child Support Agency or the Home Office. We recommend that there be clear official guidelines for the use of DNA testing for child support and immigration control purposes, and that these should reflect the concerns about the intrusiveness and potential consequences for family members of DNA parentage testing. In our view, such guidelines should state that DNA testing should only be used in situations where no other evidence is available.
10.27	Private paternity testing, such as that commissioned for civil court cases, may fall under a voluntary UK Code of Practice on Genetic Paternity Testing Services.

	However, the Code of Practice is not legally enforceable and does not apply to paternity testing services offered by overseas providers. We recommend that the effectiveness and relevance of the Code of Practice on DNA Paternity Testing should be considered as part of our review of direct offering to the public of genetic testing services.
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List of national contacts

Austria

Höfler Gerald

University of Graz
Auenbrugger Platz 25
A-8036 GRAZ
Tel : +43 316 3858 3654
Fax : +43 316 384329
Email : gerald.hoefler@meduni-graz.at

Dr. Dietmar Vybiral

Ministry for Health and Women
Dep.IV/B/12 (Biotechnologie, Gentechnik)
Radetzkystrasse 2
A-1031 Vienna
AUSTRIA
Phone: +43-1-71100- 4486
Fax: +43-1-715 24 05
e-mail: dietmar.vybiral@bmgf.gv.at
<http://www.gentechnik.gv.at>
<http://www.bmgf.gv.at>

Belgium

Professor Jean-Jacques Cassiman

Arthur Bax and Anna Vanluffelen chair of Human Genetics
Center for Human Genetics/ Centrum Menselijke Erfelijkheid
Campus Gasthuisberg
B3000 Leuven
Tel +32 16 345860
FAX +32 16 345997
Jean-Jacques.Cassiman@med.kuleuven.ac.be
www.eurogentest.org
www.cfnetwork.be
<http://cwisdb.cc.kuleuven.ac.be/research/T/team272330.htm>

Frank Hulstraert

Frank Hulstaert, MD, MSc, FFPM
Arts-Expert
Federaal Kenniscentrum voor de Gezondheidszorg
Résidence Palace (Blok A, 10e verdieping)
Wetstraat 155
B-1040 Brussel
www.kenniscentrum.fgov.be
Tel. direct: +32 (0)2 287 33 73; secretariaat: +32 (0)2 287 33 88
Fax: +32 (0)2 287 33 85
E-mail: Frank.Hulstaert@kenniscentrum.fgov.be

Czech Republic

Assoc. Prof. Milan Macek Jr. M.D., D.Sc.

Institute of Biology and Medical Genetics
Department of Molecular Genetics - CF Center
University Hospital Motol & 2nd School of Medicine of
Charles University Prague
V Uvalu 84, Praha 5, CZ 150 06, Czech Republic
tel: + 420 224 439 548 /including voice mail/
FAX: + 420 224 433 523
Milan.Macek.Jr@LFMOTOL.CUNI.CZ
WebSite <http://camelot.lf2.cuni.cz/funkovai/ublq/cf.html>

Denmark

Axen Maja-Lisa

The Ministry of the Interior and Health
Slotsholmsgade 10-12
DK-1216 København K
tlf. + 45 33 92 32 55
fax. + 45 33 92 32 80
e-mail: mla@im.dk

P. Saugmann-Jensen, M.D.

Senior medical officer
National Board of Health
2ND Department
Islands Brygge 67
DK-2300 Copenhagen S
Phone 7222 7794 (direct line)
psj@sst.dk

Estonia

Elve Raukas, M.Sc

Scientist, molecular biologist
Molecular Diagnostic Center
Tartu University Clinics United Laboratory
Oru st. 3, Tartu
Estonia
Tel: +372 7422167
E-mail : Elve.Raukas@kliinikum.ee

Prof. Dr. Andres Metspalu

Estonian Biocenter
University of Tartu
Riia 23, EE 2400, Tartu
Estonia
tel. + 372- 7- 420 210
FAX: + 372 - 7 - 420 286
E-mail: andres@ebc.ee

Finland

Jalanko Anu

Head of Department
National Public Health Institute
Department of Molecular Medicine
Biomedicum Helsinki
P.O.B 104, 00251 Helsinki. Finland
Fax: +358-9-47448480
Tel: +358-9-47448392
E-mail: anu.jalanko@ktl.fi

France

Delpech Marc

Hopital Cochin
123, Bd Port Royal
F-75014 Paris
Fax 33(1) 58 41 15 80
Tel. 33 (1) 58 41 15 23 (secrétariat)
E-mail: delpech@mail.cochin.inserm.fr

Germany

Schnieders Birgit

Federal Ministry of Health and Social Security
Am Propsthof 78 A
53121 Bonn
Tel.: +49 - 1888 - 441 - 3137
Fax.: +49 - 1888 - 441 - 4842
Mail: birgit.schnieders@bmgs.bund.de

Mueller-Reible Clemens R.

Institut of Humangenetics
University Wuerzburg
Biozentrum, Am Hubland
D-97074 Wuerzburg
Tel: (+)49-931-888-4063
Fax: (+)49-931-888-4069
e-mail: crm@biozentrum.uni-wuerzburg.de

Ireland

Barton David E

National Centre for Medical Genetics
Our Lady's Hospital for Sick Children
Crumlin, Dublin 12, Ireland.
tel +3531 409 6749
fax +3531 456 0953
e-mail: david.barton@olhsc.ie
<http://www.genetics.ie>

Italy

Taruscio Domenica

Istituto Superiore di Sanità
Viale Regina Elena 299
00161 ROME
Tel : +39 06 4990 2805
Fax : +39 06 4938 7140
e-mail : taruscio@iss.it

Florida Giovanna

Istituto Superiore di Sanità
Viale Regina Elena 299
00161 ROME
Tel : 0039 06 49902805
fax: 0039 06 49387140
e-mail : giovanna@iss.it

Latvia

Prof. Dr. Astrida Krumina

Department of Medical Biology and Genetics
Medical Academy of Latvia, Dzirciemia 16, 1007, Riga
TEL. + 371 2450708
FAX. + 371 782 81 55
Email: astrida.krumina@rsu.lv

Lithuania

Prof. Dr. Vaidutis Kučinskas

Human Genetics Centre, Vilnius University Hospital "Santariskiu Klinikos"
Santariskiu street 2, 2021, Vilnius
tel/fax: +370-2-796 365, tel. +370-2-720 077
E-mail: Vaidutis.Kucinskas@santa.lt

Dalia Kasperaviciute

Dept. of Human and Medical Genetics
Vilnius University, Faculty of Medicine
Santariskiu str. 2
08406 Vilnius
Lithuania
tel. +370 5 2365 197
fax +370 5 2365 199
e-mail: dalia.kasperaviciute@gf.vu.lt

The Netherlands

Henriette Roscam Abbing

Prof. of health law, University of Utrecht
Legal counsellor, health law,
Ministry of health, welfare and sport
e-mail: h.roscamabbing@minvws.nl
h.roscamabbing@law.uu.nl

Portugal

Jorge Sequeiros, M.D., Ph.D.

Professor de Genetica Medica UnIGENE + CGPP - IBMC,
Univ. Porto R. Campo Alegre 823 4150-
180 Porto
Portugal
E-mail: jsequeir@ibmc.up.pt

Slovakia

Dr. Ludovít Kadási

Institute of Molecular Physiology and Genetics
Faculty of Natural Sciences, Comenius University
Mlynska Dolina B2, SK 842 15, Bratislava
tel: + 421 7 796 444; FAX. + 421 7 814 083
Email: kadasi@fns.uniba.sk

Dr. Frantisek Cisarik, PhD.

Ministry of Health
E-mail: cisgen@nspza.sk

Spain

Albert Martinez Armando

Coordinador Científico
Ministerio de la Ciencia y tecnología
Consejo Superior de Investigaciones Científicas (CSIC)
Joaquín Costa,22
MADRID
Tel : +34 91 563 54 82 ext. 320
Fax : +34 91 564 26 44
Email : aalbert@cindoc.csic.es

Sweden

Kristoffersson Ulf

Associate professor, senior consultant
Department of Clinical Genetics
University Hospital
S-221 85 Lund
tel +46-46-17-33-63
fax +46-46-13 10 61
E-mail :Ulf.Kristoffersson@klingen.lu.se

Lena Jonsson

Director, special expert, responsible for questions on medical ethics
Ministry of Health and Social Affairs
103 33 STOCKHOLM
SWEDEN
phone: +468405 3309
fax:+468781 0481
E-mail: Lena.Jonsson@social.ministry.se

United Kingdom

Dr Mark Bale

Head of Genetics
Scientific Developments and Bioethics Division
Department of Health
659C Skipton House
London Road
London SE1 6LH
UK
Tel +44 (0)20 7972 5169
Mob. 07789 653514
Fax +44 (0)20 7972 1717
E-mail: Mark.Bale@dh.gsi.gov.uk

Dr Dianne Kennard

Scientific Developments and Bioethics Division
Department of Health
653C Skipton House
London Road
London SE1 6LH - UK
Tel. 0207 972 1606
E-mail: Dianne.Kennard@dh.gsi.gov.uk