



# ETHICAL AND SOCIAL ASPECTS OF GENETIC TESTING SERVICES: ISSUES AND POSSIBLE ACTIONS

## A Technology Assessment Contribution to the EUROGENTEST Network of Excellence

Danielle Bütschi Häberlin

October 2005

*In memory of our appreciated and regretted colleague,*
*Susanna Jonas*

## Contents

## Foreword

The EUROGENTEST Network of Excellence has commissioned viWTA, the Flemish Parliamentary Technology Assessment Institute, to contribute to its first year program (2005) with a start-TA on the ethical and social aspects of genetic testing services.

In order to collect a maximum of up-to-date, essential research throughout Europe in a period of only six months, viWTA designed a stepwise interactive process. It was built around a limited number of key researchers and stakeholders from diverse backgrounds, both from inside and outside the European Parliamentary Technology Assessment (EPTA) network.

The first step consisted of a pre-framing workshop in Brussels. A limited number of problem-representative experts were asked to make a long-list of their colleagues throughout Europe. They also designed the framework of a questionnaire, which was refined by viWTA afterwards. In a second step, a broad range of experts was asked to complete the questionnaire. From the group of respondents, a balanced set of experts and stakeholders was invited to a two-day workshop, which was co-organised and hosted by the Institute for Prospective Technological Studies in Seville (IPTS). A specialist from “Timeout” facilitated this workshop.

The information gathering process was completed by two rounds of digital consultation, not only with the participants of the Seville workshop, but with the other respondents of the questionnaire as well. An independent expert reviewed the draft of the report, and his remarks were for the most part taken into account for the final report. The result is a concise, up-to-date status quaestionis, supplemented with recommendations for policy and research.

A highly experienced independent TA expert, Dr. Danielle Bütschi Häberlin, has been subcontracted by viWTA to guard the continuity of the process, and the consistency and the quality of the resulting output. She was involved from beginning to end: from the pre-framing workshop, throughout the Seville meeting to the digital consultations. This final report is her work.

By multiplying feedback and quality control loops, viWTA succeeded in staying faithful to its principles of interactivity and thorough validation of results. However, it was immediately clear that an in-depth analysis of all the varied arguments of the stakeholders was out of reach. If further information is needed, we recommend a follow-up study.

We hereby wish to acknowledge and express our gratitude to the experts that have actively shared their know-how by answering the questionnaires and/or by participating in one of the workshops and in the digital consultation. Without their valuable input and constructive interaction, this study would not have been possible. We cherish our memories of Susan Jonas, one of the experts, whose life tragically ended shortly after her participation in the process. Last but not least we want to express our gratitude for the work performed by Danielle Bütschi Häberlin. She distilled a tangible deliverable out of the creativity unleashed by the process. It

was a challenge, and together with her we're proud to present the result. The responsibility for these results is now in the hands of policymakers. The impact of this report on their work will be the ultimate test for this endeavour.

Dr. Willy Weyns

Project Manager

## Executive Summary

The project “Ethical and Social Aspects of Genetic Testing Services: Issues and Possible Actions” aims at providing expert-based knowledge on existing or new ethical and social implications of genetic testing services, in order to support decision-making processes at the research, clinical and policy levels. It is integrated within the Ethical and Legal Unit of the EUROGENTEST Network of Excellence (part of the priority “Science and Society” of the 6th EU Framework Programme), a joint initiative for developing high-quality genetic testing services throughout Europe.

The project has been placed with the Flemish Institute for Science and Technology Assessment (viWTA), which advises the Flemish Parliament on scientific and technology developments and society-related issues. As part of a large network of public and private organisations giving policy advice on implications of scientific and technological developments, viWTA can rely on a wide and interdisciplinary expertise. Consequently, it is in a position to provide the EUROGENTEST partners with valuable and comprehensive data on ethical and societal implications of genetic testing services, and with prospective insight into actions to be taken in this domain.

According to its mission and working processes, viWTA wanted the project to follow an interactive process, in which experts would provide input on the past and ongoing debate on ethical and social aspects of genetic testing services. Experts were also called upon in order to draw up recommendations for further action in the field. The project design is further explained in *Chapter 2,* but comprises basically four major phases:

1. In a first defining phase, TA specialists and key-players from the EUROGENTEST Network of Excellence were invited to take part in a “framing workshop”. The aim of this workshop was to define the scope of the project, to specify the type of projects under investigation, to identify the ethical and social aspects to be investigated and to suggest further experts to be involved in the process.
2. In a second phase, viWTA selected a sample of 18 projects to be reviewed, which all addressed ethical and social implications of genetic testing services. A questionnaire sent to the 18 concerned project managers provided valuable information for understanding the issues at stake and the stakeholders involved (see *Chapter 3*).
3. Third, based on the answers received through the questionnaires, a first overview on the ethical and social implications addressed by these projects and the stakeholders’ views was drafted. It offered a basis for discussion at a workshop of international experts co-organised and hosted by the Institute for Prospective and Technological Studies (IPTS), which took place in Seville on 26-27 May 2005. Geneticists, ethicists, social scientists, Technology Assessment and Health Technology Assessment specialists, industry representatives and patients’ representatives were invited to

complement and discuss the overview, and to make recommendations for further activities to be undertaken related to the ethical and social aspects of genetic testing services.

4. Finally, all the inputs gathered through the questionnaires and through the expert workshop in Seville were compiled in the present report, which was previously submitted electronically to all participants of the project (project managers who completed questionnaires, participants of both the framing workshop and the Seville workshop).

The main findings of the project are presented in Chapters 4 and 5. *Chapter 4* offers a synthesis of the ethical and social implications of genetic testing services, based on the responses given by selected project leaders to the questionnaire and on the discussions held at the Seville workshop. Issues are manifold: they concern the topics of consent, right (not) to know, access to genetic tests, quality of genetic testing services, privacy and confidentiality, discrimination and stigmatisation, selection and enhancement, a possible geneticisation of society, implications of commercialisation of genetic tests, and the costs-related questions. All these topics are widely debated among stakeholders, decision-makers and the general public. Some issues have been discussed for a long time (such as privacy, consent or discrimination), whereas others are emerging parallel to advances in science and technology.

*Chapter 5* proposes a list of recommendations intended to decision-makers concerned by the genetic testing services and their ethical and social implications. This list was elaborated by the experts attending the Seville workshop and contains different kinds of action (research programmes, stakeholder dialogue, public participation projects, pilot projects, etc.) able to support decision-makers. More precisely, the experts proposed the following initiatives:

- **A need for clarification of terms: Defining genetic testing services**

  There should be a clear definition of genetic data and genetic testing when investigating or debating any related issue. Moreover, to allow for comparison and harmonisation, a common definition of genetic tests should be aimed at.

- **Knowledge of underlying values and norms**

  Knowledge of values and norms which underlie positions and actions of the players (geneticists, medical personnel, researchers, patients, etc.) and the general public has to be gained in order to understand decision-making processes and to make stakeholders aware of their choices and trade-offs.

- **A better understanding of the conflicts**

  A better understanding of the in-depth positions of different interest groups involved in the debate on genetic testing services is required. Such investigations would provide a

better picture of the conflicts and reasons for divergent points of view held between groups and within the same groups.

- **Fostering public debate and participation**

  Research on public perceptions in southern European countries and new member states is needed, as well as participatory projects involving experts, politicians and citizens, In northern European countries, where public debate is far more advanced, public participation activities would need to be coordinated in order to allow cross-national comparisons.

- **Informed consent: constraints and expectations**

  The question of how to realise the principle of free and informed consent in practice is still open. There is a lack of empirical evidence on the expectations of patients or research volunteers with respect to pre-counselling, information and consent in clinical and research contexts. Empirical sociological and psychological studies are required to better understand the needs of the different players, be it the patients, the research volunteers or medical personnel.

- **Generic consent for biobanks: a new concept in need of examination**

  The legal status of generic consent needs to be investigated at a European level, as well as its practical implications. Generic consent should also be discussed within society, which implies the launching of participatory projects.

- **Persons judged incapable of giving consent: who should decide?**

  The question of persons lacking capacity to give consent requires further investigation, taking into account that no unique solution can be envisaged. Adjustments could be made according to the age of the patient (when the persons concerned are minors), the kind of diseases considered and the type of tests. Since the debate confronts diverging points of views, interested parties and experts should have the opportunity to present and discuss their positions in a constructive dialogue (stakeholder and public dialogues).

- **Values and norms: a reality-check**

  The many normative statements relating to genetic testing services should undergo a “reality-check”, based on empirical research. This reality-check would throw light upon how the general principles published in research documents or guidelines could be implemented in order to fit the expectations of the different players involved (patients, families, medical personnel and researchers). It would also provide an empirical basis for a fine-tuning between principles and practices, when necessary.

- **Studying the specificities and common factors of genetic testing services**

  The specificities of genetic tests should be studied further, as should the common factors shared with other medical fields. This would permit the application of what has been learnt from other similar medical situations, but would still allow for identification of those areas that are unique to genetic testing and to apply appropriate action. Projects should also be launched in order to reflect on the ways debates, research and recommendations addressing genetic testing services may benefit other health sectors.

- **Economic impact of genetic testing services**

  There is a clear need to obtain reliable information on the effectiveness of genetic testing services for different diseases, compared to other treatments or prevention strategies. Assessing the economic impacts of genetic tests should consider the health care system as a whole. This implies the assessment not only of the clinical effectiveness of genetic tests, but also the way social and psychological dimensions might influence the health care system in terms of delivery of services, human resources or possible side-effects on patients' and relatives' health.

- **Population screening**

  Each screening programme should be evaluated on a case-by-case basis before being implemented. Such an evaluation requires input from a broad range of evidence, which may thus imply that preliminary research and pilot projects have to be conducted. Monitoring is also recommended for ongoing programmes.

- **Genetic tests on the Internet**

  Investigations should be carried out – and regularly updated – on the kind of genetic products available on the net and the people using them. Research should also be launched in order to better understand the motivation of people to buy genetic tests. Based on these investigations, legal studies and coordination efforts should be undertaken to enforce national and international rules aimed at minimising the risks of "e-genetic tests".

- **Selection and enhancement: risk or opportunity?**

  There is an urgent need to undertake activities on the creation of designer babies through pre-implantation diagnosis, as several European countries authorize – or will be soon authorizing – this practice. Other research trends and evolution in genetics demonstrate further risks of selection, not at birth but during a person's lifetime (e.g. sport genomics). Our societies should reflect on how to cope with non-medical genetic tests and the general principles to apply before these become reality.

- **Discrimination: a reality?**

  The actual discussion on discrimination occurs mainly at a theoretical level, without reference to concrete cases. A reality-check should be undertaken about possible discriminatory practices related to genetic testing. Such investigations would certainly be worthwhile in order to put the debate on discrimination on a factual basis.

- **Ethnicity**

  Population based genetic testing and pharmacogenetics must be thoroughly studied and debated, in terms of both equity and efficiency. The question of ethnicity with respect to patenting and marketing also deserves further scrutiny. Experts call above all for a public dialogue.

- **A structured overview of European Union funded research**

  European funded projects have produced valuable results, which have to be considered when discussing the ethical and social implications of genetic testing services. A synthesis report should be launched, aiming at providing an inventory of all European funded research projects addressing in one form or another the ethical and social implications of genetic testing services.

- **Bringing EUROGENTEST to the people**

  As a network joining the major players in Europe in the field of genetic testing services and working on the improvement of genetic testing services throughout Europe, EUROGENTEST might offer a productive and necessary platform for a discussion between the public, geneticists and researchers. Participatory projects should thus be developed as an activity of EUROGENTEST.

# 1. Background and aims of the project

Over the past decades, scientific and technological developments have allowed scientists to improve their understanding of how genes work and how genes are linked to diseases. Increasingly, researchers are able to identify mutations that can lead to specific disorders. Tests for gene variations make it possible not only to detect diseases already in progress, but also to predict diseases yet to arise or (in the case of multifactorial disorders) to identify predisposition to certain conditions.

These new capabilities profoundly affect the methodologies and strategies used in medical therapy and health care, as they open up new preventive, diagnostic and therapeutic possibilities. However, they also raise critical ethical and social issues relating to the practices of geneticists (at the clinical and research level), of insurers, employers, pharmaceutical or medical industries, etc. Genetic testing services, alongside the new perspectives they provide, raise questions related to fundamental principles of our societies such as confidentiality, privacy, autonomy, protection against discrimination, informed consent etc.

## 1.1. Improving the knowledge base for decision-making

These questions and the methods of tackling them have been widely studied and discussed since the beginning of genetic testing, be it at the academic, clinical or policy level. Moreover, various ethical, professional and political bodies, at both national and international levels, have published a considerable number of guidelines and recommendations relating to these issues. The main recommendations can be traced back to national statutes and international conventions (e.g. the Convention on Human Rights and Biomedicine of the European Council), as well as to clinical practices and research protocols. All these efforts clearly show that the stakeholders involved are open and sensitive to the ethical and social issues of genetic testing.

However, after more than 15 years of reflection and discussion on the ethical and social implications of genetic testing services, some questions still remain open. Experts, geneticists, social scientists, health professionals, concerned patients, NGOs and the general public hold divergent positions on certain issues, so that the debate on the way to handle them is still going on. Moreover, new ethical and social questions regularly emerge, as scientific and technological developments in genetics open new possibilities in testing. Genetic testing thus remains an issue engendering lively discussion and concrete initiatives must be taken by stakeholders or decision-makers in order to address ethical and social-related issues.

The project “Ethical and Social Aspects of Genetic Testing Services: Issues and Possible Actions” takes its starting point from this observation. It has as its objective the provision of expert-based knowledge on existing or new ethical and social implications of genetic testing services, in order to support decision-making processes at the research, clinical and policy

levels. More precisely, different forms of expert inputs (i.e. input from project managers, from academics, from stakeholders' representatives, etc.) provide the basis for presenting the ethical and social challenges of genetic testing services and for making recommendations on possible action in order to meet these challenges.

As for its form, the project delivers a status report on the ethical and social implications of genetic testing based on the analysis of past and ongoing projects addressing these topics. This analysis was conducted with experts in the fields concerned who ultimately drew up recommendations for further initiatives to be taken to address these questions.

## 1.2. A project part of the EUROGENTEST Network of Excellence

The project "Ethical and Social Aspects of Genetic Testing Services: Issues and Possible Actions" is part of the EUROGENTEST Network of Excellence (included in the priority "Science and Society" of the 6$^{th}$ EU Framework Programme), which has as its objective to "structure, harmonise and improve the overall quality of [genetic testing] services, while paying substantial attention to issues resulting from testing including legal, health policies and health economic impact, IPR (Intellectual Property Rights), ethical and social questions: confidentiality, informed consent, employment and insurance" (see www.eurogentest.org). The project is integrated within Unit 4 of EUROGENTEST, dedicated to the ethical, legal and social issues of genetic testing services (see Eurogentest 2003). In this respect, it should provide a basis for further activities of this unit of the Network of Excellence.

## 1.3. Aims of the project

The project "Ethical and Social Aspects of Genetic Testing Services: Issues and Possible Actions" has been designed to provide the EUROGENTEST Network of Excellence with an analysis of existing gaps in the study of the ethical and social implications of genetic testing services and with concrete actions to address these gaps. The final outcome of the project is a list of recommendations on future initiatives to be taken to support decision-making on genetic testing services. These recommendations are primarily addressed to the members of the EUROGENTEST Network of Excellence, but also to other key players at regional, national or international levels from the political, scientific, medical, and health care fields and from society as a whole.

## 2. Description of the project

The EUROGENTEST Network of Excellence commissioned the Flemish Institute for Science and Technology Assessment (viWTA), which advises the Flemish Parliament on scientific and technology developments and society-related issues, to carry out the project "Ethical and Social Aspects of Genetic Testing Services: Issues and Possible Actions".

As a member of the Parliamentary Technology Assessment community, viWTA has as its mission to support decision-making processes by providing knowledge on scientific and technological advances, on their implications for society and on the role and attitudes of concerned players (see www.viwta.be). In this respect, viWTA generates both scientific and social knowledge, which will eventually be used by stakeholders and decision-makers to "shape" technologies at scientific, technological, economic, professional or policy levels.

### 2.1. A Technology Assessment perspective

In order to achieve its mission, the Flemish Institute for Science and Technology Assessment (viWTA) uses a mix of scientific and interactive instruments developed within the Technology Assessment community. *Scientific instruments* are meant to provide objective and interdisciplinary knowledge about the consequences of a technology, the relevant stakeholders, the control mechanisms needed to implement the technology, etc. *Interactive instruments,* for their part, offer dialogue platforms for stakeholders and the general public, in order to share experiences and points of views or to build a common position.

The project "Ethical and Social Aspects of Genetic Testing Services: Issues and Possible Actions" is addressed to decision-makers and stakeholders involved in the development and delivery of genetic testing services. It has been designed as a Technology Assessment process, i.e. "a scientific, interactive and communicative process which aims to contribute to the formation of public and political opinion on societal aspects of science and technology" (see Decker and Ladikas, 2004: 14). Based on this approach, the project aims at gaining interdisciplinary expert knowledge on the social and ethical implication of genetic testing services through experts' interaction.

As a matter of fact, experts are involved throughout the project, from the drawing-up phase to the detailed report. Their input is gathered through face-to-face interaction (at workshops) and written contributions (questionnaires, electronic consultation). In this respect, the project does not offer an in-depth study of the ethical and social implications of genetic testing services. Rather, it has been conceptualised to benefit from the expertise of researchers and technology assessment (TA) specialists. The outcomes of this interactive process will be distributed to EUROGENTEST's members and other decision-makers, to support them in their decision-making processes.

## 2.2. Scope of the project and research questions

The mandate given by EUROGENTEST to viWTA is to give an overview of existing projects addressing the ethical and social aspects of genetic testing services, to identify existing gaps in the field and to draw up recommendations for further action aimed at supporting decision-making processes.

According to this mandate, the project clearly focuses on ethical and social aspects of genetic testing services, leaving aside more technical issues such as the quality of tests, effectiveness of genetic screening, etc. Moreover, the report looks at overarching ethical and social issues related to genetic tests, and excludes questions related to very specific tests. Finally, the project considers genetic tests in a broad perspective, as it addresses issues related to research involving genetic tests (i.e. genetic test development or research based on genetic information), to the testing itself (as a technical/medical intervention) and to the services provided before, during and after the testing (e.g. genetic counselling).

## 2.3. Phases of the project and players

The project "Ethical and Social Aspects of Genetic Testing Services: Issues and Possible Actions" comprises several phases, which all resort to expert input according to the interactive approach used by viWTA in many of its activities. Experts have been called upon to frame the project, to elaborate an overview of projects and initiatives addressing the ethical and social issues of genetic testing services, to identify gaps in scientific and social knowledge related to these issues and to elaborate recommendations.

A team of TA practitioners at viWTA led the whole project. A professional facilitator was included in the project to lead the experts' discussions at two workshops (see below). Moreover, a senior consultant monitored the whole process, analysing and synthesising all the expert inputs in a report.

The following list presents the main steps of the project:

1. In a first defining phase, TA experts and key players from the EUROGENTEST Network of Excellence were invited to take part in a "framing workshop" in Brussels (2nd March 2005). The aim of this workshop was to define the scope of the project, to specify the type of projects under investigation, to identify the ethical and social aspects to be investigated and to suggest further experts to be involved in the subsequent steps of the project. The results of this phase are described in Appendix II.
2. In a second phase, viWTA selected a sample of 18 projects to be reviewed. These projects were selected to obtain an overview of the scope of research and concrete initiatives addressing the ethical and social implications of genetic testing services. The projects were selected using criteria permitting the choice of a broad sample of projects, which vary according to the themes addressed, their methodology and their context:

- Type of tests considered in the project (diagnostic tests, predictive tests, pharmacogenetics, screening[1])
- Moment of testing considered in the project (pre-implantation, prenatal, neonatal, minors, adults)
- Methodology of the project (scientific, participatory/interactive)
- Type of institution leading the project (academic, policy advice)
- Country or region considered (geographical diversity)

The sample was selected among 90 projects, which were either suggested by experts who participated in the framing workshop or found through an Internet search. All the projects selected share a common objective of advising policy-makers, professionals or other involved stakeholders. Even though EUROGENTEST primarily considers genetic testing services in a European perspective, a Canadian project was also included in the sample as being of particular interest (APOGEE-Net, see 3.16).

3. A questionnaire was sent to the persons who were (or had been) in charge of the projects selected. Its objective was, on the one hand, to achieve a better understanding of the aims and scope of the projects reviewed and, on the other, to have an overview of the positions and arguments on ethical and social issues raised by genetic testing services (see chapter 5). The questionnaires thus contained questions relating to the aims of the projects, the type of genetic testing services they were investigating, the ethical and social aspects they considered, the ways these aspects were discussed and the impact achieved by the projects (see Appendix III for a full version of the questionnaire).
4. These questionnaires were all returned to viWTA. Based on the answers provided, a first overview on the ethical and social implications addressed by these projects and the stakeholders' views was drafted.
5. This overview offered a basis for discussion at a workshop of international experts, co-organised and hosted by the Institute for Prospective Technological Studies (IPTS), which took place in Seville on 26-27 May 2005. 16 Experts from the fields of parliamentary Technology Assessment, Health Technology Assessment, ethics and sociology, as well as representatives of bio-industries, insurances and patients' organisations were invited to complement and discuss this overview, and to make recommendations for further activities to be undertaken related to the ethical and social aspects of genetic testing services (see Appendix IV).
6. All the inputs gathered through the questionnaires and the expert workshop in Seville were compiled in a draft report, which was submitted electronically to all participants of the project (project managers who completed questionnaires, participants of both the framing workshop and the Seville workshop). The experts who participated in the

[1] During the framing workshop, experts also mentioned nutrogenomics as an issue worth considering in the sample, but no completed project addressing the ethical and social implications of nutrogenomics could be identified.

Seville workshop had already received the proceedings of their discussions on which they could react. The draft report therefore contained an amended version of the recommendations incorporating their comments.

7. All the comments and proposals received from this last (and electronically) interactive phase were then included in a final report. Remarks resulting from an external review process by an independent expert have also been integrated in this final round.

## 2.4. Limits of the project

The final report synthesising all inputs gathered throughout the project does not pretend to offer a consensual view on the existing research and discussions on genetic testing services or on action to be taken in this field. This would go far beyond the objective of the project. Analysis and recommendations presented in the report should be considered as the result of a cross-fertilization exercise, where experts from different backgrounds could share their knowledge, analysis, concerns and wishes with respect to genetic testing services. This is the reason why some of the recommendations might appear contradictory or, at least, incompatible. The aim of the project is to highlight some paths for action for stakeholders and decision-makers. These are not meant as “ready-to-use” solutions, but rather as “food for thought” which needs to be further elaborated and discussed.

Moreover, even though viWTA involved experts from different sectors and with various backgrounds, the project cannot pretend to present an exhaustive and detailed analysis of the ethical and social implications of genetic testing services. Nor does it present an in-depth analysis of the argumentation lines and of the existing tensions among or between the existing opinions. This would go far beyond the scope of a project meant to present a status report of issues and possible actions. Accordingly, the present report offers an overview of the main challenges with which decision-makers are (or will in the near future) be confronted, and the positions different stakeholders take about them.

## 3. A sample of projects on the ethical and social implications of genetic testing

The ethical and social issues of genetic testing services have been addressed in many projects, both at national and international levels. Academic, clinical or policy bodies have addressed these issues in many different settings, e.g. ethical research, socio-psychological research on certain types of diseases, ethical or professional committees leading to the elaboration of guidelines or regulation, public involvement procedures, etc.

A sample of 18 projects has been selected which covers both the thematic and geographic diversity (for more details on the selection procedure, see section 2.3 above). The projects were selected by viWTA using several sources: an Internet search, proposals from participants in the framing workshop and from members of the parliamentary Technology Assessment Network (EPTA Network). The projects are of a diverse nature, from academic research to public dialogue or guidelines. Regardless of their academic or practice oriented nature, they all produced results able to support decision-making processes on genetic testing services.

The following pages briefly present the 18 projects selected. For each project, a short summary is provided, together with basic information obtained through the questionnaire.

### *1) Predictive genetic research: where are we going? (1995)*

*Rathenau Institute (Platform of Science and Ethics), The Netherlands*

The Rathenau Institute has as its objective the support of social and political opinion making on issues arising from technological and scientific developments. Via the organisation of various activities, it stimulates social and political debate over a wide range of subjects. The project "predictive genetic research" was one of the first public debates organised by the Institute. Based on prior expert-workshops and on a background study on the topic, a so-called consensus conference[2] has been organised in order to allow a group of citizens to discuss this topic and the related issues with experts and ethicists. This conference led to a final statement written by the involved citizens, which has been presented to the Dutch Parliament.

**Type of genetic tests:** Predictive test: late onset disease; Predictive test: predisposition; Screening of population

**Time of testing:** Prenatal, Minors

**Field of application:** Health

**Testing initiative:** Medical personnel, Individual initiative, Public authorities, Insurers

[2] The participatory method of consensus conferences intends to facilitate the participation of large segments of society in the on-going discussions on technology choices. It was developed by the Danish Board of Technology in the late 1980s, and since then has been applied for varying issues worldwide (see Joss and Durant 1995).

**Methodology:** Literature analysis, Expert workshop, Participatory method (with citizens, patient groups, experts, stakeholders, politicians)

More about the project:
"Predictive genetic research, where are we going?" Rathenau report, April 1995.

### *2) EUROSCREEN I and II (1993 – 2000)*

*University of Central Lancashire, Centre for Professional Ethics, United Kingdom*

Euroscreen I examined genetic screening and testing in Europe with particular reference to predictive medicine, with subgroups on childhood testing; historical precedents; and concepts of health and disease. Euroscreen II examined genetic screening with special reference to insurance, commercialisation and public awareness, including the experimental Gene Shop at Manchester Airport.

**Type of genetic tests:** Diagnostic test: clinical diagnosis; Diagnostic test: defining diseases; Predictive test: late onset disease; Predictive test: predisposition; Screening.

**Time of testing:** Pre-implantation; Prenatal; Neonatal; Minors; Adults

**Field of application:** Health, Paternity; Family planning

**Testing initiative:** Medical personnel; Individual initiative; Public authorities; Employer; Insurer; Commercial organisation.

**Methodology:** Literature analysis; Expert workshop; Survey; Participatory method (involving citizens, patient groups and experts)

More about the project:
Chadwick et al. 1997; Chadwick et al. 1999; Levitt 2000.

### *3) Status and perspective of genetic testing (2000)*

*Office of Technology Assessment at the German Parliament (TAB), Germany*

The project gives an overview of the achievements of genome research and its perspectives. The project considers both the technological issues and the social and policy issues. In this respect, the overview comprises questions related human genetic counselling and diagnosis, prenatal diagnosis, preimplantation diagnosis, genetic tests in employment, gene-tests for insurance and regulatory aspects.

**Type of genetic tests:** Diagnostic test: defining diseases; Predictive test: late onset disease; Predictive test: predisposition; Pharmacogenetics; Screening of populations.

**Time of testing:** Prenatal, Neonatal, Adults

**Field of application:** Health; Family planning; Workplace; Insurance contracts.

**Testing initiative:** Medical personnel, Individual initiative; Commercial organisation

**Methodology:** Literature analysis; Expert workshop; Survey.

More about the project: Hennen et al. 2000.

### 4) *Citizens' Conference on Genetic Testing (2001)*

*Stiftung Deutsches Hygiene-Museum, Germany*

Upon invitation from the Deutsches Hygiene-Museum, a group of 19 randomly chosen citizens from all over the Federal Republic of Germany and of a broad age range and from a variety of occupational backgrounds, came together in Dresden to form an opinion on the issue of genetic testing. It was the first nationwide citizens' conference on genetic testing and was based on the model of "consensus conferences" developed by the Danish Board of Technology.

**Type of genetic tests:** Diagnostic test: defining diseases; Predictive test: late onset disease; Predictive test: predisposition

**Time of testing:** Pre-implantation; Prenatal, Adults

**Field of application:** Health, Quality of life, Family planning

**Testing initiative:** -

**Methodology:** Participatory method (involving citizens and experts)

More about the project:
http://www.buergerkonferenz.de/pages/start_en2.htm

### 5) *Testing our genes (2002)*

*The Danish Board of Technology, Denmark*

The Danish Board of Technology has been set up in order to promote ongoing discussion about technology, to evaluate technology and to advise the Danish Parliament and other governmental bodies in matters pertaining to technology. In the project "Testing our genes", the ethical, societal and economical consequences of genetic testing services have been discussed by lay people, experts, and politicians on a consensus conference.

**Type of genetic tests:** Diagnostic test: defining diseases; Predictive test: late onset disease; Predictive test: predisposition; Pharmacogenetics, Screening.

**Time of testing:** all

**Field of application:** Health; Family planning.

**Testing initiative:** Medical personnel; Individual initiative.

**Methodology:** Participatory method (involving citizens, patient groups, experts, stakeholders)

More about the project:
http://www.tekno.dk/subpage.php3?article=496&language=uk&category=11&toppic=kategori11

6) ***Predictive Genetic Testing for Hereditary Breast and Ovarian Cancer and Colorectal Cancer. An Assessment (2002)***

*The Institute of Technology Assessment - ITA (Austria Academy of Sciences), Austria*

The aim of this project is to analyse the current scientific knowledge and the situation of genetic counselling on predictive genetic testing for hereditary breast and colorectal cancer. It is based on non-systematic literature research using several databases. In addition to the scientific basis on genetic testing, the different diagnostic test methods, the benefit of early detection methods and prophylactic interventions are described and analysed in the context of individual/familial and social consequences.

**Type of genetic tests:** Diagnostic test: clinical diagnosis; Predictive test: late onset disease; Predictive test: predisposition; Screening of populations.

**Time of testing:** Minors; Adults

**Field of application:** Health; Quality of life; Family planning

**Testing initiative:** Medical personnel; Employer, Insurer; Commercial organisation.

**Methodology:** Literature analysis; Participatory method (involving patient groups, experts and stakeholders); Expert workshop; Interviews.

More about the project:
Jonas et al. 2002 and http://www.oeaw.ac.at/ita/ebene5/d2-2b21.pdf

7) ***Preimplantation diagnostics practices and statutory regulation in seven selected countries (2003)***

*Office of Technology Assessment at the German Parliament (TAB), Germany*

The project is based on the analysis of case studies from other countries (Belgium, Denmark, France, UK, Italy, Norway and Italy). It has been designed in order to compare statutory regulation and practical use of pre-implantation genetic diagnostics (PGD), to achieve a better understanding of the relationship between different regulatory models and the evolution of supply and demand in the use of PGD. The study included both countries with comparatively restrictive regulation of preimplantation diagnosis and countries where there is no legal regulation or the existing statutory framework permits PGD without establishing individual indications or prerequisites for examining embryos in particular cases.

**Type of genetic tests:** Predictive test: late onset disease; Predictive test: predisposition

**Time of testing:** Pre-implantation

**Field of application:** Health; Family planning

**Testing initiative:** Medical personnel; Individual initiative

**Methodology:** Literature analysis; Survey.

More about the project:
Hennen and Sauter 2004.

### 8) *Initiative on genetic testing (2003-2004)*

*King Baudoin Foundation, Belgium*

The King Baudoin Foundation launched, in 2003, a large initiative on genetic testing. This initiative was divided into three closely related projects: (1) Expert conference "Testing of human beings"; (2) Public Forum "Is it in my genes?", based on the consensus conference model developed by the Danish Board of Technology; (3) Support of projects stemming for the civil society, schools and Universities ("Dialogues in Society in the use of genetic testing": 15 projects supported).

**Type of genetic tests:** Diagnostic test: clinical diagnosis; Diagnostic test: defining diseases; Predictive test: late onset disease; Predictive test: predisposition; Screening.

**Time of testing:** Pre-implantation; Prenatal, Neonatal, Minors, Adults

**Field of application:** Health, Quality of life, Forensic, Paternity; Family planning

**Testing initiative:** Medical personnel; Individual initiative; Public authorities; Employer; Insurer; Commercial organisation.

**Methodology:** Literature analysis; Expert workshop; Participatory method (involving citizens, patient groups, experts and stakeholders).

More about the project:
http://www.mesgenes.be/

### 9) *Expert group on ethical, legal, social aspects of genetic testing (2004)*

*European commission DG research, Science and society*

An Expert Group was invited by the European Commission to discuss the ethical, legal and social aspects of genetic testing and to publish recommendations. ("25 Recommendations on the ethical, legal and social implications of genetic testing") It included representatives from the industry that produces or uses genetic tests, from NGOs (in particular, patient organisations with clear interests in the subject), and scientists and representatives from academic institutions with different backgrounds (law, philosophy, ethics, and medicine).

**Type of genetic tests:** Diagnostic test: clinical diagnosis; Diagnostic test: defining diseases; Predictive test: late onset disease; Predictive test: predisposition; Pharmacogenetics; Screening.

**Time of testing:** Preimplantation; prenatal, Neonatal, Minors, Adults

**Field of application:** Health, Family planning

**Testing initiative:** Medical personnel; Public authorities; Employer; Commercial organisation.

**Methodology:** Literature analysis; Expert workshop; Consultation of experts.

More about the project:
European Commission 2004.

***10) Pharmacogenetics and Pharmacogenomics (2004)***

*Swiss Centre for Technology Assessment (TA-SWISS), Switzerland*

TA-SWISS mission is to provide the public and the political authorities with high-quality and unbiased information on new technologies. The project "Pharmacogenetics and Pharmacogenomics" first gives a review of facts in summarising the state of the art of biomedical issues, the point of view of societal groups, media coverage, public opinion and the legal situation. In a second part, the project reviews options for further developments and assesses the advantages and the risks considering aspects related to medicine, economy, public health, society, ethics, and law. The report concludes with a series of recommendations concerning legislation, research, data protection, insurances, professional education and public debate.

**Type of genetic tests:** Pharmacogenetics

**Time of testing:** Adults

**Field of application:** Health

**Testing initiative:** Pharmaceutical companies; Diagnostic laboratories.

**Methodology:** Literature analysis; Survey; Expert interviews.

More about the project:
Rippe et al. 2004.

***11) Children (not) allowed? Ethical aspects of the genetic testing in children (2003-2004)***

*Center for Biomedical Ethics and Law, Faculty of Medicine (K.U. Leuven), Belgium*

This research project aims to study the normative and ethical challenges of genetic testing in minors for carrier tests and predictive genetic tests.

**Type of genetic tests:** Predictive test: late onset disease; Predictive test: predisposition; Carrier tests.

**Time of testing:** Minors

**Field of application:** Health; Family planning

**Testing initiative:** Individual initiative; Parents

**Methodology:** Participatory method (involving citizens and experts)

More about the project:
Borry et al. 2005

***12) Models of genetic analysis for Cystic Fibrosis (2004)***

*Commissione di Studio sulle modalità di analisi genetica per Fibrosi Cistica, Gruppo di Studio per la Fibrosi Cistica della Società Italiana di Pediatria, Italy*

The project consists in the elaboration of a document aiming at giving indications on the type of Cystic Fibrosis genetic tests to be implemented for different kind of categories of users.

**Type of genetic tests:** Diagnostic test: clinical diagnosis; Screening; Carrier testing.

**Time of testing:** Prenatal, Adults

**Field of application:** Family planning

**Testing initiative:** Medical personnel

**Methodology:** Literature analysis; Expert workshop.

More about the project:
http://www.pnlg.it

***13) Biobanks career (1) and A sociological approach of informed consent for genetic epidemiology research (2) (parallel projects) (2005)***

*(1) INSERM; Toulouse; CNRS CIRUS, Toulouse and (2) CNRS CERTOP, Toulouse (France)*

The project on sociological analysis on informed consent in genetic epidemiology (2) examines through interviews, the attitude of participants to a genetic epidemiology research involving a biobank (for a study on cardiovascular multifactorial disease risk factors including genetic factors). Different patterns of attitudes that show the discrepancy between the theoretical norm and the participants attitudes are examined.

The project on biobanks careers (1) is a study involving legal and sociological approaches addressing the evolving status of samples, data and biobanks through an empirical analysis of cases of different biobanks at different moment of their developments.

**Type of genetic tests:** Predictive test: predisposition; Biobanks.

**Time of testing:** Adults

**Field of application:** Health; Biomedical research

**Testing initiative:** Medical personnel; Researchers.

**Methodology:** Literature analysis; Survey; Interviews; Observation; Participatory method (involving stakeholders)

More about the project:
(1) Ducournau 2004, 2005 and (2) Cambon-Thomsen 2003, 2004, Hirtzlini et al. 2003.

***14) Multidisciplinarity in counselling for predictive testing: a professional and societal challenge (1998 – 2005)***

*University Hospital K.U.Leuven (Psychosocial Genetics Unit and Clinical Genetics Unit), Belgium*

The project focuses on predictive testing for two hereditary cancers: hereditary breast/ovarian cancer (HBOC) and hereditary non polyposis colorectal cancer (HNPCC). In both cases carriers of the mutation can take preventive measures. The research, imbedded in the daily practice of the genetic clinic, is aimed at getting more insight in the psychological aspects of predictive testing for HBOC and HNPCC. The project ends up with recommendations for practice.

**Type of genetic tests:** Predictive test: late onset disease

**Time of testing:** Adults

**Field of application:** Health; Family planning

**Testing initiative:** Individual initiative

**Methodology:** Literature analysis; Interviews and questionnaires.

More about the project:
http://www.kuleuven.be/psychogen

***15) Ethics in Medical Genetics (ongoing project)***

*University of Nijmegen (Centre for Society and Genomics), The Netherlands*

The project "Ethics in Medical Genetics" is a workpackage of the INES project (The Institutionalisation of ethics in science policy), a co-ordinated action of the 6th framework programme of the European Union. The aim of Workpackage IV Ethics in Medical Genetics is to provide a comparative overview of current ethical practice and to assess what infrastructure would be needed to deal with certain applications of medical genetics in the future. Two case studies on cascade screening for familial hypercholesterolaemia (FH) and pre-employment screening serve to explore current practice as well as future options, with focus on the normative aspects.

**Type of genetic tests:** Predictive test: late onset disease; Predictive test: predisposition; Screening

**Time of testing:** Adults

**Field of application:** Health, Pre-employment genetic testing

**Testing initiative:** Individual initiative; Public authorities; Employer

**Methodology:** Expert workshop

More about the project:
http://www.cesagen.lancs.ac.uk/research/related/ines.htm and
http://www.society-genomics.nl/?page=407

***16) APOGEE-Net, a Network to support policy making in genetics (ongoing project)***

*A project funded by the Canadian Institutes for Health Research (CIHR), Canada*

APOGEE-Net, an interdisciplinary capacity enhancement network funded for 5 years by the CIHR, brings together the producers, disseminators and users of knowledge (including citizens) to support policy making in the area of genetics. Knowledge transfer, transdisciplinary research and capacity building are key objectives of the network. The proximal outcomes of this project should be the production of relevant policy oriented research in the area of genetics, research capacity building, consolidation of the network itself, and the progressive refinement of an innovative knowledge transfer strategy as a result of a formal evaluation process built into the network activities. The expected distal outcomes are increased use of research data in policy making, receptor capacity building,

informed public participation to the debate, better planning and management of genetic services, and the development of a sustainable technology transfer in genetics.

**Type of genetic tests:** Diagnostic test: clinical diagnosis; Predictive test: late onset disease; Predictive test: predisposition; Screening.

**Time of testing:** -

**Field of application:** Health; Quality of life; Family planning

**Testing initiative:** -

**Methodology:** Literature analysis; Expert workshop; Participatory method (involving experts, stakeholders and researchers from advisory bodies)

More about the project:
http://www.aetmis.gouv.qc.ca/en/mod.php?mod=userpage&menu=39&page_id=122

***17) Human Genetic Databases: Towards a global ethical framework (ongoing project)***

*University of Geneva (Bioethics Institute) and World Health Organization (Department of Ethics, Trade, Human Rights and Health Law), Switzerland*

The project explores the conditions under which genetic databases may be established, kept, and used in ethically acceptable ways that are based on the principles of fairness, equity and respect for human rights. The project unfolds in several steps: review of the scientific literature, comparative analysis of existing normative frameworks and interviews. The aim of the project is to produce practice guidelines and to produce a training module for researchers and members of research ethics committees worldwide.

**Type of genetic tests:** Screening; Population-based genetic databases and data collections.

**Time of testing:** Minors; Adults

**Field of application:** Health; Research

**Testing initiative:** Medical personnel; Commercial organisation; Research organisation.

**Methodology:** Literature analysis; Survey

More about the project:
http://www.who.int/ethics/topics/hgdb/en/

***18) Psychosocial effects of molecular genetic diagnosis: the case of X-linked learning disabilities (ongoing project)***

*Centre for family research, University of Cambridge, United Kingdom*

The project is a complementary study to the GOLD Study (the Genetics of Learning Disability), which will try to find mutations in samples of DNA collected from families where there are individuals affected by significant learning disabilities. The project will explore what it is like to take part in GOLD, and the social impact for family members if molecular genetic diagnosis becomes available.

| | |
|---|---|
| **Type of genetic tests:** | Diagnostic test: clinical diagnosis; Diagnostic test: defining diseases. |
| **Time of testing:** | Prenatal, Minors; Adults |
| **Field of application:** | Health; Quality of life; Family planning |
| **Testing initiative:** | Medical personnel; Individual initiative |
| **Methodology:** | Survey with patients |

More about the project:
http://www.wellcome.ac.uk/assets/wtx024800.pdf

# 4. Ethical and social issues of genetic testing services: an overview

Shortly after the framing workshop, all the contact persons of the projects selected received a questionnaire to complete to provide input material for the expert workshop in Seville. As well as questions aimed at describing the aims and content of the project, they were asked to specify the issues addressed in their projects (closed question, with possibility for the respondents to add new categories). A recapitulation of their answers is presented in section 4.1.

Then, for each issue noted, respondents were asked to specify which positions and arguments came into play during their project, the degree of consensus or dissent and the key players involved. Their replies were summarized in a first overview, which was presented to the experts attending the Seville workshop. This workshop was a crucial phase of the project, as the invited experts were asked to discuss the current ethical and social challenges of genetic testing services and to elaborate recommendations intended to support decision-makers (see section 3.2 and Appendix IV). Based on this, a synthesis of the ethical and social aspects of genetic testing services was elaborated (see section 4.2).

## 4.1. Aspects / issues considered in the projects

Table 1 gives a first overview of the issues considered in the different projects of our sample. We see from this table that most projects reviewed consider a broad range of ethical and social issues associated with genetic testing services and do not restrict themselves to particular questions. This can, of course, be explained by the choice of projects included, which were selected as being somehow representative of the current activities in the field and for their contribution to the general discussion on genetic testing services. But this feature also shows that ethical and social aspects are all linked and require a comprehensive approach. For instance, privacy aspects cannot be separated from discussions on consent. Similarly, discrimination and stigmatisation fears are closely linked to the debate on the geneticisation of society.

Even if the limited number of projects reviewed does not permit any quantitative analysis, it is evident that some issues are covered in nearly all projects, whereas others appear in only some. For instance, the right to know, free and informed consent and accessibility to genetic tests have been considered in 14 out of 18 projects, whereas costs to the health sector appear in only four projects. These figures certainly provide insights into the strength of the academic and social discussions on the issues, but we should in no way extrapolate the importance of the issues.

***Table 1:*** *Projects and addressed issues*

| | Right to know (or not to know) | Free and informed consent | Privacy | Ownership + control over results and genetic material | Discrimination | Accessibility to genetic tests | Geneticisation of society | Commercialisation of genetic services | Costs to the health sector | Quality of genetic testing services | Religion | Other |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1. Predictive genetic research (Rathenau) | X | X | X | X | X | X | | X | | | | |
| 2. Citizens' Conference on Genetic Testing (Deutsches Hygiene-Museum, Dresden) | X | X | X | X | X | X | X | X | | X | | |
| 3. Testing our genes (Danish Board of Technology) | X | X | X | X | X | X | X | X | X | X | X | |
| 4. Preimplantation diagnostics practices and statutory regulation (TAB) | X | | | | | X | | X | | X | | Regulation State control |
| 5. Initiative on genetic testing (king Baudoin Foundation) | X | X | X | X | X | X | | X | | X | | |
| 6. Expert group on ethical, legal, social aspects of genetic testing | X | X | X | X | X | X | X | X | X | X | | Information; Media; Dialogue |
| 7. Biobanks careers + Informed consent for genetic epidemiology research (France) | | X | X | X | | | | | | | | |
| 8. EUROSCREEN I and II | X | X | X | X | X | X | X | X | | X | | |
| 9. Multidisciplinarity in counselling for predictive testing (University Hospital K.U.Leuven) | X | X | X | X | X | X | | | | X | | |
| 10. Status and perspective of genetic testing (TAB) | X | X | | | X | | X | X | | X | | |

| | Right to know (or not to know) | Free and informed consent | Privacy | Ownership + control over results and genetic material | Discrimination | Accessibility to genetic tests | Geneticisation of society | Commercialisation of genetic services | Costs to the health sector | Quality of genetic testing services | Religion | Other |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 11. Children (not) allowed? (K.U. Leuven) | X | X | X | X | X | X | | | | | | |
| 12. Ethics in Medical Genetics (University of Nijmegen) | X | | X | X | | X | | | | | | |
| 13. Predictive genetic testing for hereditary breast, ovarian cancer and colorectal cancer (ITA) | X | X | X | X | X | X | X | X | X | X | | Information and media |
| 14. Models of genetic analysis for Cystic Fibrosis (Italy) | | | | | | X | X | X | | X | | |
| 15. Pharmacogenetics and Pharmacogenomics (TA-SWISS) | X | X | X | | X | | | | | | | |
| 16. APOGEE-Net (Canada) | | | | | | X | | | X | X | | |
| 17. Human Genetic Databases (WHO and University of Geneva) | X | X | X | X | | | | X | | | | |
| 18. Psychosocial effects of molecular genetic diagnosis | | X | | X | | X | X | | | X | | |
| **Total** | **14** | **14** | **13** | **13** | **11** | **14** | **8** | **11** | **4** | **12** | **1** | |

## 4.2. Issues, positions, arguments and players

The following section offers a synthesis of the ethical and social implications of genetic testing services which were discussed at the Seville workshop, based on the responses given by selected project leaders to the questionnaire. For each issue, the critical questions are presented, as well as the arguments involved and, wherever possible, the positions of experts, professionals, patients, relatives, research volunteers and of the general public. Whenever possible, the references of the projects in which these questions have been addressed are mentioned. However, many issues and arguments were addressed during the Seville workshop without reference to a concrete project, so that the synthesis only partially refers to concrete projects. Moreover, some discussions remained at a general level, which has been reflected in the overview.

As a matter of fact, the synthesis should not be viewed as an exhaustive and comprehensive report on the ethical and social implications of genetic testing services. It summarises the inputs received from selected projects and invited experts. However, projects and experts were chosen to cover a wide variety of disciplines, practices, points of view and topics. We can thus consider that it reflects the current state of the debate and indicates the major ethical and social challenges facing genetic testing services.

### 4.2.1. Freedom of choice

Freedom of choice appears to be a basic principle pertaining to genetic testing, be it in the clinical or research context. All stakeholders, observers and decision-makers involved in genetic testing services seem to agree that free and informed consent has to be guaranteed, whatever type of genetic test is being considered. For instance, the European Commission, in its “25 recommendations” states, “medically relevant genetic testing should never be imposed and should always be a matter of choice” (European Commission 2004: 12 - project 9). In its guidelines, the European Commission also highlights the importance of informed consent in research based on genetic testing. Despite this, the way to guarantee freedom of choice remains open to debate. In particular, the topics of counselling, information and socio-economic pressures are considered as still critical. The form of consent is also emerging as a new discussion topic among experts and interest groups.

#### *Consent*

It is widely acknowledged that any genetic test should not be passed without the patient’s free and informed consent. This point of view is particularly widespread among the general public, as we can read in many consensus conferences final statements such as “The principle of individual autonomy and control of one’s own body should be firmly maintained in our society

(Danish consensus conference - project 5) or “There should not be any mandatory screening of populations, except for special health reasons” (Consensus conference in Germany - project 4).

However, recent empirical studies show that expressing free and informed consent is not that simple. For instance, in a recent French sociological study, research volunteers passing genetic tests declared that they just want to trust their doctors. For the majority, signing the form caused doubts and questioning. In actual fact informed consent was considered as a way for doctors to protect themselves and to escape their responsibilities (Ducournau 2004 and 2005). These empirical results are rather unexpected, but they prove that the debate on free and informed consent is far from being closed.

Besides this, questions are still open with respect to the consent of persons unable to take decisions (minors or persons with intellectual impairment). Whereas most national legislations imply that families (i.e. the legal respondents) should consent on behalf of people with intellectual impairment, some interest groups and professionals demand that the person tested should be consulted when possible (see project 18). The same kind of debate occurs with minors. Even though the law clearly states that legal respondents have to decide on behalf of minors, some groups or professionals consider that from a certain age, children and teenagers are in a position to give their consent on whether or not to pass a genetic test and should thus at least be consulted (see Borry et al. 2005 - project 11).

As noted by experts taking part in the Seville workshop, genetic research and the related development of biobanks shed new light on the issue of consent. Biobanks allow for storage of samples of human bodily substances (e.g. cells, tissue, blood, or DNA) that are or can be associated with personal data and information on their donors In this respect, they constitute an important resource for identifying the causes and mechanisms of a large number of diseases. For many researchers and specialists, however, their potentialities are limited by a strict concept of consent. As a matter of fact, specific consent as it is currently implemented implies asking the donors for consent for any new use of their material or data, which some consider could impose limits on research. In order to guarantee research, most existing regulations on biobanks acknowledge that explicit donor consent can be dispensed with subject to certain conditions, such as the anonymisation of samples and of data, or the restriction to a certain time limit or type of research. But this remains an unsatisfactory solution as samples and data are restricted to certain uses. Recently, some voices called for the introduction of generic consent, where donors would give their consent for any research use of their genetic information. These claims obviously receive a certain interest from decision-makers, especially in the United Kingdom as the recent Tissue Act gives legality to generic consent.

### *Counselling*

It is widely acknowledged that a precondition for consent in the clinical context is the existence of an unbiased pre-test counselling, where patients are informed about the test itself, the kind of

results it will deliver and the way to interpret them. Counselling is especially important for predictive tests, as they deliver uncertain information as to whether and when the disease will occur (see project 6).

Most actors consider that the issue of consent deserves special attention. Only few take unbiased counselling as being realised. As a matter of fact, most professionals consider that even though the regulatory framework for pre-test counselling is available, the concrete implementation of counselling is still confronted with practical difficulties. They regret that in many cases, resources are lacking to offer systematic and comprehensive pre-counselling services (see project 6).

Citizens who have taken part in consensus conferences are much more critical towards counselling. They share the fears and concerns of the professionals with respect to the lack of funding for genetic counselling. But they also consider that current standards of counselling are insufficient (see Danish consensus conference - project 5). And in some cases, they even call for governments to establish more formal rules ensuring the conditions for free choice (see Belgian consensus conference, project 8)[3].

Moreover, whereas most stakeholders and the general public trust genetic testing centres to offer high-quality counselling as long as certain conditions are met, some critical groups (such as feminist groups, see project 3) call for the possibility for patients to ask for second opinions and alternative counselling. These groups want to avoid any monopoly of doctors and demand that independent institutions and initiatives be supported in order to offer patients alternative counselling. Even though most stakeholders agree that there are a variety of routes to obtain support, there is no majority demand for the formal establishment of alternative counselling.

Finally, some experts in social sciences and ethics voice more fundamental criticism. They cast doubts on the possibility of ensuring qualified counselling in the future if test possibilities continue to steadily develop and testing becomes so simple and cheap that non-specialists can use them (see Hennen et al. 2000 - project 3).

***Information and education***

Freedom of choice occurs not only through adequate counselling. According to many stakeholders, it also implies that the public receives objective and up-to-date information, so that individuals can really make an enlightened choice when offered the possibility of passing genetic tests. Some citizens taking part in a consensus conferences, for instance, declared that without possessing basic knowledge on genetics, patients might be strongly intimidated by their doctors, which is of course an obstacle to free and informed consent (see for instance consensus conferences in Belgium - project 8 and in Germany - project 4).

[3] These demands have been in part taken into consideration by the Swiss government, as its new law on genetic tests declares that for predictive genetic tests, counselling prior to the testing is mandatory.

***Socio-economic pressure***

Social or economic pressures must also be considered when addressing the issue of freedom of choice. This aspect is especially put forward by citizens taking part in consensus conferences or by critical organisations. For instance, participants of the Dutch consensus conference (project 1) noted that social and family pressure might force individuals into taking a genetic test. The line of reasoning behind this position is that representation of the illness, handicap and care possibilities may put subjective limits on freedom of choice. This is especially the case for prenatal testing, but also when offering the test to persons with a family history of specific disease. Screening programmes may also increase the social pressure towards testing. Some experts attending the Seville workshop noted that in certain countries, this is already the case for neonatal screening, as neonatal genetic testing is automatically offered to parents. Even though they can refuse it, the future parents do not really have the opportunity to choose, as it has become standard practice. Moreover, the time pressure under which decisions must be made often constrains freedom of choice. The same concerns have been voiced by the citizens who participated in the German consensus conference for prenatal genetic testing (project 4), as they state "we are deeply concerned that prenatal testing has spread so much over the last few years… The problem is that too many women resort to prenatal diagnosis without careful consideration".

Another concern relates to genetic testing at workplaces. In the Dutch and German consensus conferences (projects 1 and 4), for instance, citizens expressed their concern about people being forced to accept testing at the workplace if no legislation restricts the use of genetic tests by employers on their employees. They fear that the financial and economic consequences of refusing to pass a test may limit the freedom of choice of the workers. This position is also held by trade unions, which fear that workers will have to adapt to the workplace, whereas the tendency until now has been to adapt the workplace to the health and security of the workers (see Hennen et al. 2000 - project 3). But trade unions, as highlighted at the Seville workshop, do not call for a total ban of genetic tests at the workplace, as forbidding all tests by employers might endanger the safety of some workers (especially those working in environments at risk).

### 4.2.2. The right to know (or not to know)

In the discussion on the ethical and social implications of clinical genetic testing, the right to know and the right not to know are often presented as important principles pertaining to medical practice. The debate on the right to know (or not to know) has, however, to be distinguished, according to whether we take the patient's or relative's perspective. As a matter of fact, getting to know (or not know) about a genetic condition has varying implications according to whether patients or their relatives are concerned.

***The right to know (or not to know) of patients***

The right to know implies that each person is entitled to obtain genetic information related to his present or future health status when it can be made available through genetic tests. In this respect, the right to know appears as a crucial right of patients. But should the right to know be applied in every case? And what about persons who do not want to know?

For some affected groups and patients' relatives groups, there should be a right not to know when no therapy is available (see project 3). Citizens who took part in the Danish consensus conference in 2002 (project 5) went even further, as they stated that no genetic tests should be offered when no treatment is available. This view is also shared by the participants of the Dutch consensus conferences who discussed prenatal diagnosis: according to them, prenatal tests should be offered only in case of illnesses for which a reasonable therapy is available (project 1). Such opinions are contested by those who consider that even though no treatment is available, it can be a relief for certain persons to obtain a diagnosis or, in case of predictive tests, to be in a position to take crucial life choices or to make changes to lifestyle in order to reduce risk (see project 5).

Beyond these disputes on the opportunity to know or not about one's genetic information, most stakeholders actually consider that both the right to know and the right not to know have to be acknowledged. The first appears as an essential patient's right pertaining to medical practice, whereas the second relates to the previous discussion on freedom of choice. The European expert group, which elaborated the EU recommendations on the ethical, legal and social implications of genetic testing recognises this, as it states that "it is imperative to recognise both the right to know and the right not to know" (see European Commission 2004: 12, project 9). Nevertheless, some experts cast doubts on the practicability of the right to know, especially when genetic testing is proposed within a therapeutic context. This point has been highlighted in the TA Study on pharmacogenetics presented in our sample (Rippe et al. 2004 - project 10): if the patient is willing to have a certain therapy but claims the right not to know, he would thus reject the therapy.

***The right to know (or not to know) of relatives***

When considering the right to know or not know, the perspective of relatives comes also into play since genetic tests give information which also concerns them. When discussing this, citizens participating in the Danish consensus conference considered that the relatives of genetically tested people should be protected from unwanted knowledge about their own genetic makeup (project 5). This point of view is also shared by many experts and professionals.

Nevertheless, respecting the relative's right not to know can place the person tested before an unbearable dilemma: he must respect on the one hand the will of his relatives, but knows on the

other hand that his relatives might have a certain condition. The citizens taking part in the Danish consensus conference have noted this difficulty. However, as highlighted at the Seville workshop, some experts consider this whole debate as purely theoretical. They consider that the relative's right to know is impracticable: according to them, it is impossible to decide not to know without knowing what there is to know.

What makes the discussion even more difficult is that the right to know of relatives has also to be acknowledged, but this might be in contradiction with privacy rights of patients (see section 4.2.5). Clearly informing a relative about a possible hereditary risk implies revealing one's own condition. Should individuals with a genetic disorder be obliged to inform their relatives about the fact that they (possibly) bear the same condition? What occurs when somebody does not want to inform his relatives? Is it the role of medical doctors to inform them, thus breaking medical secrecy? Is a doctor or a patient liable for not telling relatives important information about their health?

These questions are extremely sensitive: the rights of the one can endanger the rights of the other. This dilemma can be addressed in different ways, and we find a variety of solutions in the various national legislations. In France, for instance, a recent law states that medical doctors are not allowed to transmit genetic information to patients' relatives. They must nevertheless inform their patients about the importance of transmitting such information to their relatives, but in no case can a patient be sued for not transmitting the information. This is a clear example of a regulation where the patients' rights prevail over the right to know of relatives. In Switzerland, on the contrary, the new legislation on genetic analysis allows for doctors to transmit genetic information to relatives should a patient refuse to inform them about their condition. But they must first get the authorization of an official commission. Here, the right to know of relatives prevails over the patient's rights. In the Netherlands, a similar perspective has been chosen, as patients consulting for hereditary cancers give their authorisation on a consent form that the geneticist will inform their family members of their risk, unless they do it themselves.

### 4.2.3. Access to genetic tests

Most key players – including citizens – consider that genetic tests should be accessible to anyone, independent of status, revenue or place of residence. In this respect, genetic testing services are expected to be part of an efficient and accessible health care system. This position, however, does not mean that genetic testing services must be available without constraints and control. Basic conditions such as proper genetic counselling and proper medical indication have to be fulfilled, so that the problems and risks inherent to genetic tests (such as the way to handle uncertain information, the consequences of tests for relatives, etc.) can be kept to a minimum.

***Economic access***

It seems largely accepted that testing must be affordable and be kept affordable (see for instance the Belgian consensus conference, project 8). Otherwise, new inequities could emerge in societies. There is also a wide consensus on the fact that public insurance (social security) should pay for the tests, if the scientific evidence is valid and an improvement of outcome (reduction of mortality and/or morbidity) is demonstrated (see project 6).

Public consultations in the form of consensus conferences showed some concerns from citizens towards patents, which are considered as potential barriers to tests. This was especially the case in Belgium and Denmark, where citizens taking part in consensus conference feared that patenting might limit access to genetic tests. Professionals also share these concerns and the debate on gene patenting is currently very animated in Europe (see also section 4.2.9). Geneticists and their interest organisations (such as the European Society of Human Genetics) are not opposed to patents as such, but to their anti-social use, i.e. the creation of a monopoly by asserting rights over the gene sequence and its mutations. They thus claim that the delivery of compulsory licensing should permit other firms to make use of the license content under specified terms. Bio industries, on the contrary, consider gene patenting as a necessary instrument for financing their research efforts.

***Geographic access***

Most key players recommend that rules of access and quality standards should be uniform from one region to another, or one country to another. There should be no regional variability as to whether or not the individuals/families concerned are offered genetic testing. This view is also shared by citizens taking part in consensus conferences (see Danish consensus conference, project 5). This claim is at the core of the EUROGENTEST Network of Excellence, as this European-wide project strives to harmonize genetic testing throughout the member States.

***Information and trust as conditions for accessibility***

According to experts who participated in the Seville workshop, accessibility to genetic testing is not only limited by financial or geographical barriers. Psychological barriers have also to be considered when speaking of accessibility. These should be kept to a minimum in, for instance, ensuring confidentiality. In other words, trust towards genetic testing has to be built, otherwise patients may refuse to take a genetic test as they fear misuse. Looking at the results of consensus conferences, it is clear that the public has a cautious attitude towards genetic testing and that many latent fears are present. Providing information is certainly a good way to reassure the public, but, according to the Seville experts, building trust is much more important.

***The role of first-line doctors***

The role of first-line doctors (general practitioners and gynaecologists) is an important issue when addressing the accessibility of genetic testing services. Studies have showed some important variations with respect to their knowledge on genetics and testing, so that their role in providing a gateway into specialist care varies from country to country (see Harris 1997). For most experts, as well as for the citizens taking part in consensus conferences, their role should be expanded and genetic testing centres should contribute to this by providing first-line doctors with more information and training (see e.g. Dutch and Belgian consensus conferences, projects 1 and 8). They would thus be in a better position to inform their patients about the availability of genetic tests, about the prevention and treatment possibilities offered and about the ways to interpret and handle the results.

Knowledge of first-line doctors on the potentialities and limits of genetic tests is especially important for predictive tests (and will become more and more important as predictive tests may increase in the future), as these tests provide uncertain information as to whether and when a disease may appear. Similarly, prenatal tests confront future parents with important ethical and psychological questions, and it is thus essential to inform and train gynaecologists about the scope and consequences of such tests (see Hennen et al. 2000, project 3).

### 4.2.4. Quality of genetic testing services

Genetic tests are based on high-level scientific knowledge, but have been developed as a technically simple and easy instrument. This can lead to obvious quality gaps, as the technology can be used by persons not possessing the medical skills to provide high quality services and an adequate psychosocial support to patients.

***Medical quality***

For most stakeholders, only qualified doctors should conduct genetic tests. According to this position, only University Hospitals can ensure the quality of testing, so that they alone should be entitled to deliver genetic testing services (see project 3). Citizens taking part in consensus conference, who put strong emphasis on the existence of a medical indication and on comprehensive counselling, share this view. In the German consensus conference (project 3), citizens even recommended that a primary, competent authority should safeguard the quality of the genetic tests by regulating the activities of doctors in the provision of these tests. Some EU countries have recognized this claim as their laws restrict the delivery of genetic testing services to University centres.

Experts at the Seville workshop noted that critical voices towards genetic testing – such as feminist groups – demand that patients and their representatives be included in discussions

relating to the provision of genetic testing services. Their views should be taken into consideration, as well as those of specialists. Such claims go in the direction of patient's empowerment, but still meet with strong resistance from medical doctors and hospital administrations.

***Quality of counselling***

Information before, during and after the testing is considered as an essential feature of quality, when considering genetic testing services from an ethical or social perspective. As a matter of fact, the issue of genetic counselling has been addressed in many projects included in the sample considered in this overview, and experts participating in the Seville workshop also stressed its importance.

According to a majority view, in order to ensure quality in counselling, multidisciplinary teams should be built up, including geneticists and doctors specialised in the condition concerned (for instance cardiologists, neurologists). Citizens taking part in consensus conferences, as well as social scientists, consider that multidisciplinarity should be insisted upon, so that counselling teams include psychologists and sociologists (see for instance the Danish consensus conference - project 5 and project 14). In Belgium, citizens taking part in the consensus conference even proposed that patients or representatives of patient associations be included in the counselling (project 8).

Moreover, professionals and observers, as well as citizens taking part in consensus conferences, deplore that in many cases, counselling is delivered without clear procedures and patterns. They thus call for the establishment of special training programmes for counsellors. In the Danish consensus conference, citizens even recommended that counsellors should hold some kind of authorization (project 5). The German citizens taking part in a consensus conference also shared this view, as they stated that there is a need for state regulation of provisions of genetic tests concerning doctors and/or counsellors (project 4).

Finally, the type of information delivered to the patients (or research volunteers) is of particular relevance to ensure quality counselling. However, experts who took part in the Seville workshop noted that it is extremely difficult to find out how to inform patients or research volunteers taking a genetic test. More information does not necessarily equate best information. Moreover, individuals may have varying needs: whereas some only want to trust their doctors, others need a detailed explanation of the test and its implications. Resulting from this situation and findings, some stakeholders claim the right for patients to define the level of information they should receive. Others insist on the fact that patients should not only be informed about the test itself, but also about secondary information that the test may generate (for instance information on paternity). This claim is especially voiced with respect to pharmacogenetics, as some tests can be used not only to determine how a person reacts to a particular drug, but also whether that person is susceptible to genetic disorders (see project 10).

### 4.2.5. Privacy and confidentiality

Genetic tests deliver very sensitive information about a person's health and traits and most projects addressing the ethical and social issues of genetic testing services have as one of their central question the privacy issue. Discussions turn on the specificity of genetic information with regard to other medical data and on the control over genetic results. Moreover, some recent developments in genetic testing (especially pharmacogenetics and biobanks) raise special privacy concerns.

#### *Is genetic information specific?*

Many professionals, researchers and industry representatives (especially insurance representatives), consider that genetic material and information should be treated no differently than other biomedical samples and data. Existing rules protecting medical information are sufficient to protect patients (or, in the case of research, donors) from any misuse of their genetic samples and data.

This view is opposed by interest groups (such as citizens' rights advocacy) and by some professionals (geneticists, counsellors), who demand restrictive rules of data protection for genetic information. According to them, the results of genetic tests deliver much more information on patients and their relatives than do other medical tests. Without any strict privacy rules, insurers or employers could obtain genetic information and eventually make a selection on the basis of the test results (see also section 4.2.6). Citizens throughout Europe also share this view, as the consensus conferences considered for our overview show (projects 1, 4, 5 and 8). Based on these concerns, many stakeholders and experts call for anonymous genetic sample and data collection, as a basic condition for building trust towards genetic research.

In fact, experts attending the Seville workshop considered that the whole issue of privacy requires a clear definition of what is understood by genetic information. Much confusion and conflict in the debate arise from ambiguous or diverging concepts of genetic information. These clarification efforts concern equally public discussion and professional dispute.

#### *Ownership and control over results*

Looking at the discussions held in consensus conferences, it is clear that for most citizens, genetic information belongs to individuals and should be protected from any third parties due to its sensitive nature. If this information is transmitted, it should always be rendered anonymous (see for instance the German and Danish consensus conferences - projects 4 and 5). This position implies that any possible use of genetic information may occur only with the person's express consent. Professionals and scientists largely share this position, as long as the discussion turns on the transmission of genetic data to private organisations (such as insurers

and employers) or to private research institutes. With respect to this latter point, experts who participated at the Seville workshop noted that the frontier between public and private research might not be so easy to draw. As a matter of fact, many test providers are private (but publicly ruled) and their results might be of interest for public research. Moreover, some of them noted that a strict application of express consent in the research context limits scientific and technological developments in genetics.

***Pharmacogenetics-related issues***

Privacy is an important issue related to the development of pharmacogenetic tests, as a widespread use of "made-to-measure drugs" will imply knowledge about genetic traits of the patient, which could lead to a dissemination of genetic information and endanger patients' privacy. However, according to certain experts, there is no reason to worry about privacy and confidentiality, as pharmacogenetic information is related to drug metabolism and not to diseases. Thus, it does not affect a patient's life to the same extent as, for instance, predictive genetic testing. However, all experts do not share this optimistic view. A recent Technology Assessment study highlighted conflicting visions on this topic (Rippe et al. 2004, project 10). In particular, the study refers to the concerns of certain experts that secondary information relating to disease could be obtained through pharmacogenetic tests. They thus demand that restrictive privacy rules should be adopted also in this domain.

***Biobanks and genetic research related issues***

Parallel to the increasing constitution of biobanks for research purposes, the debate about protection of genetic samples and data is an issue which raises many concerns. Only few States have a well-developed legal framework in this domain and there is certainly a need to gain more knowledge on the privacy issues raised by biobanks, on the possible measures to be implemented in this respect, and on the various points of view concerning these issues and the proposed solutions (see project 17).

Considering the appreciation delivered by the experts during the Seville workshop, there is no evidence of a consensual view on the level of data protection to be achieved for genetic research and in biobanks. Whereas some call for strict statutory data protection provisions and anonymous samples, others consider that too strict a regulation might raise barriers to research and should thus be prevented. Here, the issue of generic consent (see section 4.2.1) is presented as a possible solution that might reconcile both privacy concerns and research interests.

### 4.2.6. Discrimination and stigmatisation

Besides privacy concerns, fears of discrimination and stigmatisation occupy an important place in the ethical and social debate on genetic testing. Most ethical guidelines, legislation and recommendation papers address this issue in one form or another.

#### *Stigmatisation of groups*

Despite the many recommendations about the topic, fears of stigmatisation remain very vivid, especially by persons (or parents of persons) carrying a certain condition. For instance, as stated by experts at the Seville workshop, organisations of affected persons or parents claim that some groups are indeed discriminated against through the fact that genetic tests are offered for the condition they have. They feel that society wants to get rid of the condition – and of them!

Ethicists and ethnologists also point out risks of racial and ethnic discrimination, mainly related to screening programmes carried out on certain ethnic groups characterized by a high occurrence of a certain condition. Moreover, as noted during the Seville workshop, genetic screening may reinforce existing discrimination in certain societies. For instance, gender discrimination could come into play in the case of a condition held and transmitted by women.

From a completely different perspective, the Seville experts noted that the lack of genetic tests due, for instance, to a lack of research funding, could also be considered as discrimination. This may be the case for persons affected by orphan, as they may be excluded from any treatment because of a lack of funding.

Interestingly, whereas the different consensus conferences held in various national settings showed that citizens were rather critical towards genetic testing (or at least critical with respect to some aspects), citizens do not express major fears about some groups being discriminated against or stigmatised. They remain aware of the risk and call for measures against it (such as public education and information in the Danish consensus conference – project 5), but they do not use this argument to question genetic testing, as do some critical organisations.

#### *Discrimination by insurers*

Possible use of genetic tests by insurers to discriminate against people considered to be a “bad risk” seems to raise much more concern in the general public than stigmatisation of groups based on their condition. Citizens taking part in the German consensus conference (project 4), for instance, noted that insurance companies may have a great interest in genetic tests: on the one hand because of the cost-cutting potential through facilitated care, on the other hand through the possible introduction of graduated rates based on genetic risk potential for disability and life insurance. They fear that the principle of solidarity would be undermined if insurance companies used genetic testing or had access to genetic records. These concerns are also

shared by citizens taking part in other consensus conferences in other countries, such as the Dutch, who fear that some groups of people will become uninsurable in private insurance systems (project 5).

The perspective of insurers is of a different nature. Insurers stress that the health insurance component of social security policy has to provide the same (basic) compensation for all, independent of their personal (genetic) risks. Therefore there is no danger involved in making genetic data available for this type of insurance. For private (life) insurance, the situation is somewhat different, as premiums are calculated on the basis of the insured person's risk profile. Thus, the insurer has the right to be aware of genetic results if the insured person has done such tests. This is not discrimination but a means of providing a fair risk calculation. This is especially important as the solidarity principle may be compromised if policyholders use to their advantage genetic information (i.e. if they insure a risk they know is high without informing the insurance company). Nevertheless, in no case can insurers claim the right to demand their clients pass a genetic test before signing a contract.

The political debate on this subject is very lively all around Europe. For instance, when the Swiss Parliament elaborated the recent legislation on genetic testing, most of the political debate turned around the possibility given to private life insurance providers to ask for existing genetic results. Interestingly, however, the general public seems open to consider the point of view of insurers. For instance, the participants of the Danish consensus conference recommended the development of new insurance models which should allow an even playing-field for the insured and the insurer, so that both economic and ethical criteria can be met (project 5). Similarly, in the Netherlands, the citizens called for research to be undertaken to find out ways to cover the risks of those considered as uninsurable (project 1).

***Discrimination by employers***

Parallel to the fears of discrimination by insurers, citizens are opposed to any demand by employers to require genetic information from their workers. They fear that employers could deny jobs to individuals at risk for certain diseases. Citizens taking part in the German consensus conference, for instance, claimed that "genetic test would give the employer the possibility to hire only healthy, able bodied employees" and, further, "an employee should always be given the choice to take any such test connected with his or her job". In the Netherlands, too, citizens refuse any wide use of genetic tests by employers. As stated by the participants of the Dutch consensus, "for appointment examinations, only demands [for genetic testing] that are specific to the function should be applied".

Experts attending the Seville workshop highlighted that these fears seem to be unjustified. There is no intention by employers to implement genetic testing in their human resource strategies. Even trade unions are quite confident on that matter. In some special cases,

however, where workers may be at risk or have important security responsibilities, the use of genetic tests could be envisaged, but under strict conditions.

***Pharmacogenetics***

The expected developments in pharmacogenomics may give rise to a new form of discrimination, as pharmacogenetic tests may exclude certain groups of patients ("orphan genotypes") from a given pharmacological treatment. All experts, however, have not the same position as to whether this situation has to be considered as discriminatory or not (see Rippe et al. 2004 - project 10).

On the one hand, some ethicists consider that even though there is a moral obligation to provide the best treatment available, there is no moral right to demand the development of a new drug. In this respect, orphan genotypes are not excluded from therapy, but they do not respond to current drugs. The problem is thus caused by drug discovery and not by genetic testing. Other ethicists, however, are worried about the discrimination potential of pharmacogenomics. They fear that some groups may be favoured against other groups, as some treatments may function only for a minority (cf. Nuffield Council 2003).

### 4.2.7. Selection

The potentials of genetic tests lie far beyond conventional medical tools, as they can be used in order to select individuals at their conception (pre-implantation genetic testing) or to select individuals along genetic traits. These potentials raise important ethical questions and debates, which vary according to whether the issue is about preventing the conception of children for certain medical indications (negative selection), choosing a child according to genetic characteristics (positive selection) or selecting individuals with certain genetic traits to, for instance, enhance their performances.

***Negative selection***

The issues raised by negative selection are closely linked to the debate about pre-implantation tests (PID). The debate has been very animated in most European countries. Whereas professionals and families concerned consider pre-implantation genetic testing as a legitimate alternative to prenatal screening and subsequent abortion, religious and feminist groups are strongly opposed to PID, which they consider as being a manipulation of life. For them, PID should be banned. Similarly, citizens fear that administration of PID would lead to selecting individuals using criteria such as health/illness or wanted/unwanted and would have negative consequences for the sick and disabled in our society (see e.g. the German consensus conference, project 4). But citizens are ready to allow PID in the case of severe genetic illness or high genetic risk.

Some specialists, patients and relatives, however, hold a far more liberal position. According to them, no restriction should be made on the availability of pre-implantation test. The decision to undergo PID is an individual choice and it is not for society to decide whether it is good or not.

How can we deal with these two conflicting positions? In many countries, the debate is still under way. However, most stakeholders consider that PID can assist parents with adverse genetic traits in responsible family planning. But recent legislation passed in some European countries shows that differences and disagreements remain on how PID is considered (see Hennen and Sauter 2004, project 7). Whereas some countries allow the use of PID in very restrictive cases for severe genetically caused disease (e.g. Denmark, France), others have adopted a far more permissive view including a broad range of medical indications (e.g. Belgium).

***Positive selection***

Most stakeholders and the general public largely oppose to the use of genetic testing to select embryos according to genetic traits. The prospect of choosing the sex or any other trait of a future child is far away from European cultural tenets. But opinions start to diverge when the debate moves to the issue of “saviour-siblings”, i.e. the conception of children compatible with their sick sibling.

Actually, the real debate around positive selection has been raised by pre-selection of embryos which could potentially save ill siblings. This is a comparatively new issue, which is emerging in most European countries as a hot topic. Opponents of such practices, argue mainly that the use of PID in this context is a manipulation of life. Proponents argue that the future child could save his/her sibling and this would not affect his/her own physical health, nor jeopardise his/her chance of a normal life.

As the debate is rather recent, there is little indication on the opinion of the general public about such practices.

***Selection as an enhancement tool***

Experts attending the Seville workshop noted that even though this is not yet a reality, genetic tests might be used in the future to select certain groups with respect to specific traits. This might be the case in sports, where promising athletes would not only be selected on their performances but also on their genetic predispositions. For many, this sounds like science fiction. But the Seville experts recommended that the issue of sports genomics has to be addressed in good time, since medicine is playing an ever-increasing role in sport.

In the employment sector, too, genetic information could be used in order to hire only healthy, able-bodied employees so as to reduce sick-days and cut down on costs. This prospect is strongly opposed by most stakeholders and the general public, as such practices would weaken the employee's position. A person's genetic makeup is a natural fact (up to now) and cannot be individually influenced.

### 4.2.8. Geneticisation of society

Critical voices from the social sciences and NGOs, as well as some medical experts, claim that genetic tests open the path to a geneticisation of society (see projects 3 and 6). According to them, everything that can be tested will be tested, whether or not this is rational on health care grounds. Handicaps or diverging genetic traits will be increasingly seen as an illness and the process of reproduction will be shifted from the bedroom to the laboratory! They also fear that exaggerating the weight given to genetics could lead to disproportion in public health interventions and in research funding.

For many experts, however, these trends are not specifically related to genetic testing. As highlighted at the Seville workshop, what is at stake can be viewed as a medicalisation of society, not a geneticisation of society. In this respect, the phenomenon is much more profound and complex.

Concerned patients or their families do not understand these arguments, since genetic testing offers them diagnosis and therapy possibilities. They refuse such normative statements about a geneticisation or a medicalisation of society, as they are propounded by people who do not experience the difficulties of living with a genetic disorder or with persons affected by such a disorder (see project 18).

### 4.2.9. Commercialisation of genetic tests

As with many other medical techniques, issues around the commercialisation of genetic tests raise concerns among specialists and the general public. These relate to the commercialisation of innovations (e.g. patents) and the selling of tests to the public (e.g. selling over the counter).

#### *Patents*

This issue has given rise to many discussions in policy and experts circles, especially after the patenting claims for BRCA1 and BRCA2 by Myriad. There seems to be a fundamental opposition from the professionals and the public to a “privatisation” of the human genome through patents. Citizens taking part in consensus conferences all over Europe clearly showed their opposition to a monopolisation of genetic tests resulting from patents (see for instance the Belgium and Danish consensus conferences). Similarly, recent positions of the European Society of Human Genetics clearly state their opposition to the monopoly claims of companies on gene sequences and their mutation (such as the monopoly that Myriad claimed). Concerned companies, on the other hand, consider patenting as an essential instrument able to guarantee a return on their research investments.

***Commercialisation over the counter***

In most European countries, only accredited laboratories and hospitals are authorized to deliver genetic tests, which is seen by health professionals as an important condition for ensuring adequate counselling and quality of the results. Nevertheless, in certain countries, some tests can be purchased over the counter, a situation which raises ethical and social concerns from specialists and among the public. These concerns are sharpened by the development of Internet and the possibility it opens to make genetic tests and drugs available beyond national borders.

For many stakeholders, commercialisation of genetic tests over the counter should be subject to strict regulation, or even be banned. Citizens taking part in consensus conferences also share this view. For instance, participants of the German consensus conference on genetic tests (project 4) claimed that it should not be legal to sell genetic tests or practice in private laboratories without offering proper counselling at the same time. In Denmark, citizens demanded that anybody offering genetic testing should hold an authorization and that it should not be legal to sell genetic tests without offering proper counselling (project 5). Moreover, they called for establishing control procedures for the commercialisation of genetic tests on Internet.

### 4.2.10. Costs of genetic testing services from an ethical and social perspective

Costs of genetic testing services for the health care system are primarily of an economic nature, but they also contain social and ethical dimensions, as economic constraints may affect priority settings, the type of tests offered, the accompanying measures such as counselling, etc.

***Costs to the health sector***

General assumptions such as “genetic tests will increase prevention and reduce the costs of treatments” are regularly heard when discussing the economic aspects of genetic testing services. Such statements are often made by genetic tests proponents or enthusiasts. It is also a view shared by many of the general public.

But the situation is not that simple. Experts attending the Seville workshop noted that very few data are available to give an opinion on the costs of genetic tests to the health sector. Moreover, the economic impact of genetic testing services may vary from case to case. If testing may allow for prevention and avoid expensive treatment in some cases, in other cases it may result in an increase in health costs as the detection of a condition may imply a costly therapy. And in other cases, such as prenatal tests, the cost-saving argument may prevail. In this respect, it is certainly too early to make clear and comprehensive statements on the economic implications of genetic tests, if it is possible at all. In any case, it would be exaggerated to present genetic testing as a technique able to decrease the overall costs of the

health sector. Moreover, such a vision poses some fundamental problems, as it implies that genetic testing services would be assessed only on the basis of economic considerations, setting aside ethical and psychological consideration.

The economic argument is often used for justifying the implementation of screening programmes. Such programmes are presented as promising instruments to reduce health costs: by detecting some widespread condition in the population or in specific groups, it is expected that treatment costs will decrease. However, ethicists who worked on the issue came to the conclusion that expected economic profit should never overrule free choice, a position which condemns any tentative to make genetic screening mandatory for economic reasons.

***The costs of counselling***

Most actors agree on the necessity to offer patients genetic counselling services before, during and after testing, as highlighted in many of the projects considered for this overview. However, counselling is cost-intensive and existing pressures on health costs may put genetic counselling at risk (see project 6). Professionals (especially counsellors) express such fears, as their claims for additional means for counselling remain too often unanswered. Citizens taking part in consensus conferences also share this view, as for example the Danes who call for “extra resources … to improve counselling” (project 5).

***Priority setting***

Research funding is a scarce resource, and some critical voices express their concern that genetic research is being favoured over other research. As highlighted during the Seville workshop, some claim that there is a ”biotech hype” and that other medical domains suffer from this as decision-makers privilege genetic research.

Even though it is clear that some research areas are being set aside (for instance research on orphan diseases), other experts are not concerned about genetic research being privileged. Funding invested in this medical area is proportional to its promise and other research areas also benefit from large funding sources.

# 5. Recommendations for further action

The two-day Seville workshop was structured according to two distinct parts. The first day was mainly dedicated to the evaluation and assessment of the current issues relating to genetic testing services, and to the key players involved and their related opinions. These discussions produced basic material for the synthesis on ethical and social aspects of genetic testing services presented in the previous section. On the second day, the experts were invited to develop recommendations for further action, based on their previous discussions. Their task was to propose different kinds of initiatives (research programmes, stakeholder dialogue, public participation projects, pilot projects, etc.) able to support decision-makers.

The present section reviews these various recommendations. It describes the different gaps identified by the experts, as well as the type of activities to be launched. Recommendations are listed thematically: no order of priority or ranking should be deduced from the presentation. Moreover, as the Seville workshop was designed as a brainstorming session rather than a mediation exercise, the recommendations listed should not be understood as a consensual result offering a set of complementary and coherent measures. Nevertheless, all recommendations were considered by experts as legitimate, so that the following review reflects at least a consensual view on the paths to be further investigated and discussed by decision-makers.

## 5.1. A need for clarification of terms

### 5.1.1. Defining genetic testing services

*Background*

Often, when genetic testing services and their social and ethical implications are discussed, the debate becomes quite confused since genetic testing and genetic data are considered without being clearly defined beforehand. Several definitions of a genetic test can be found in existing guidelines, recommendations or legislation. The term can refer to DNA-based tests, or it can include tests of gene products, such as proteins and metabolites, chromosomes and even acquired somatic cell mutations such as those associated with cancer.

Moreover, genetic testing services imply a wide range of tests and contexts of utilization, which also need to be adequately defined. Gene tests are different with regard to what information is delivered, what can be achieved with the test results and whether information given by genetic testing is different from information given by other conventional diagnostics.

*Initiatives to be taken*

In this respect, and as a first and general recommendation for further activities, the experts at the Seville workshop considered that there should be a clear definition of genetic data and genetic testing when investigating or debating any related issue. Moreover, to allow for comparison and harmonisation, a common definition of genetic tests should be aimed at. Other bodies, such as the European Commission independent expert group, which published the "25 recommendations on genetic testing", have actually already formulated similar recommendations[4]. For the experts at the Seville workshop, this call for clarification and common definitions is still valid and should be considered as a prerequisite for any project addressing genetic testing services.

In order to avoid any confusion, it is also important to clearly distinguish between different contexts of application and different quality of information provided by the tests (monogenetic vs. multifactor diseases, testing of actual status of health vs. predictive testing, predictive testing with a certain prognosis vs. testing with probabilistic information).

*Players*

All organisations, institutions and individuals involved in the study, communication and dialogue on the ethical, social and governance aspects of genetic testing services.

## 5.2. Understanding values, opinions and conflicts

### 5.2.1. Underlying values and norms

*Background*

Many players are involved in the use and development of genetic testing: governments, scientific institutions, medical doctors, health specialists, industries, patients and their organisations, as well as the general public. Their decisions and perceptions frame the practices pertaining in this particular medical domain, influence priorities within the health sector or for research, or set the legislative context for genetic testing services to cite but a few examples.

All these stakeholders have underlying values and norms, which will influence their choices. The ways in which they consider illness, health, privacy, autonomy and other life or social

[4] In the first recommendation of their report, the EC expert group stated that "any official statement or position should refer precisely to an explicit definition of the terms used or topic addressed; a consensus definition of genetic testing should be developed globally by all respective public and private bodies involved; the European Commission should consider taking the initiative on this topic" (European Commission 2004).

principles will affect research priorities, funding for given treatments or prevention programmes, legislative frameworks, clinical practices, etc.

*Initiatives to be taken*

According to experts present at the Seville workshop, knowledge of underlying values and norms has to be gained in order to understand decision-making processes and to make stakeholders aware of their choices and trade-offs. Consequently, they advocate research analysing the norms and values involved in decision-making on genetic testing services, which implies as a prior condition to understand who takes part in which decision. For instance, what underlying values come into play when a particular research area is given priority over another and who are the involved stakeholders? Why are certain diseases considered severe, requiring pre-implantation diagnosis whereas others are considered an acceptable risk? Why do some countries have more rigorous legal frameworks than others? What are the values and norms of doctors applying genetic tests? In-depth analysis and comparisons of the underlying values and norms would offer valuable information on the ways in which religious, social and cultural values affect perceptions of genetic testing and the practices of stakeholders. This would be an essential step to a better understanding and would acknowledge the national or regional differences existing within Europe, as well as in other parts of the globe. Knowledge of (and research on) stakeholders' perspectives can indeed be useful in situations where conflict resolution is necessary, but also more generally in preparation for participatory projects including the public (see section 5.2.3).

The experts participating at the Seville workshop also stressed that the study of values and preferences has to be complemented by the analysis of trade-offs. Decisions are the results of trade-offs and compromises between positions held by different groups in order to discover a compromise supported by the majority. But decisions can also imply trade-offs between contradicting values held by the same group or by an individual. Knowledge about how these trade-offs are made by citizens and by patients and their families can be of use to decision-makers.

Besides analysis and studies on the underlying values and norms pertaining to decision-making in genetic testing, experts suggested that concrete projects be developed in order to support decision-makers in their choices. For instance, it has been suggested that decision support systems be developed, asking for precise and considered reasons for taking up, for instance, research on specific diseases and not on others.

Experts also stressed the importance of communicating about the analysis and their results in an appropriate form, adapted to the audience (policy-makers, managers, professionals, patient organisations, citizens). In this respect, publishing results in specialised medical, ethics or sociology journals may well miss the point. Organising conferences and workshops where

stakeholders would meet with ethicists and sociologists could be a much more effective means to address and to cast light on underlying values and norms.

*Players*

Social scientists, ethicists, ethic commissions, patient organisations, religious groups and other NGOs.

### 5.2.2. A better understanding of the conflicts

*Background*

Many stakeholders intervene in the debate around genetic testing services, either for or against. However, a clear picture of who holds which position with which arguments is still lacking. A multitude of disseminated stakeholders take specific positions according to the interests they represent and no clear overall picture emerges. Moreover, genetic testing raises many questions, which vary according to the kind of disease or the type of test under consideration. Thus, some groups, which at first glance could be expected to share similar views on a given topic, will hold diverging opinions, as they are not affected in the same way by genetic testing services or because their members hold varying fundamental norms and values. For instance, parent or patient organisations, although they have in common concern for the (direct or indirect) distress caused by a severe disease, may hold different views on the question of consent or data protection. Similarly, patient groups can have diverging opinions on genetic testing depending on whether they suffer from curable or incurable disease.

*Initiatives to be taken*

What are the reasons for these diverging points of view and how to cope with them? According to experts taking part in the Seville workshop, a better understanding of the in-depth positions of different interest groups involved in the debate on genetic testing services is required. Such investigations would provide a better picture of the conflicts and reasons for divergent points of view (especially ethnic and cultural variations) held between players and within the same groups. This is an essential step in any effort aimed at solving the existing conflicts or at mediating between opposing positions. The aim, however, is not to eliminate diverging views in favour of a common opinion (this would be a naïve and impracticable objective), but to reveal routes which take into consideration the diverging views and interests of the players and stakeholders involved, so that each feels recognized. This is a very demanding objective, but it appears the only way to at least partially defuse conflicts.

*Players*

Social scientists, ethicists, concerned persons (patients, parents, families), stakeholders.

### 5.2.3. Fostering public debate and participation

*Background*

Most ethical guidelines acknowledge the need for collaboration between stakeholders and for public consultation. For instance, the European Commission, in its "25 recommendations" states that "an opportunity for public dialogue between different stakeholders [should] be organised, offering participants equal opportunities for expression" (European Commission 2004).

*Initiatives to be taken*

At the European level, a great deal of information is available about public perceptions in the northern European countries since many surveys and public dialogue initiatives have been implemented in these countries. However, little is known about the public perceptions in the new member states and southern Europe. According to the experts at the Seville workshop, research on public perceptions in these countries is needed, as well as participatory projects involving experts, politicians and citizens.

In northern European countries where public debate is far more advanced, public participation activities would need to be coordinated in order to allow cross-national comparisons. As a matter of fact, all participatory projects relating to genetic testing services were carried out on a national basis: the issues discussed were mainly based on national debate, with very few recommendations relating to more global governance aspects. As a consequence, even though these participatory projects all addressed a common topic (genetic tests), it is very difficult to compare their recommendations in a European perspective. For this reason, some participants at the Seville workshop suggested that dialogue projects on a supra-national level, involving several European countries, should be developed. Such a process is currently under way with a Europe-wide debate on brain science (Meeting of Minds[5]). More such projects – experimenting with methods and institutionalisation – are needed in order to gain concrete impressions of public perception Europe-wide, and genetic services would be an obvious theme for this.

On a more abstract level, experts at the Seville workshop suggested that the content and methodology of participatory projects involving citizens should be compared. With regard to content, they would be interested in comparing the content of public discussions to that of academic literature in terms of production of original solutions, priorities, trade-offs and compromises. With regard to method, the experts were aware that there is already quite an extensive literature on citizen participation. Nevertheless, some specific methodological aspects certainly warrant special attention, such as the nature of information sharing at the outset of these projects, the kind of support to decision-making that these participatory procedures are

[5] Meeting of Minds: European Citizens' Deliberation on Brain Science, http://www.meetingmindseurope.org.

best suited for (e.g. support for decision-making in research, clinical practices, health care delivery or regulation) and how such initiatives can be bridged with other types of action (research programmes, stakeholder dialogues, pilot project, etc.).

Some experts also called for involving the public in concrete research projects. The idea is not to ask citizens to decide on the type of research to conduct, but to integrate them in the reflections on the framework of research. Together with the ethics committees or similar bodies, their role would be to discuss whether the proposed research programmes meet ethical standards with respect to basic principles such as data protection, freedom of choice, autonomy, etc.

*Players*

Technology assessment institutions, science museums and other similar independent organisations.

## 5.3. From principles to practice

### 5.3.1. Informed consent: constraints and expectations

*Background*

The principle that genetic testing has to be done on a free and informed consent basis is widely accepted. However, in practice, this is far from obvious. We learned from the projects considered in the viWTA sample that some volunteers who enrolled in epidemiology research preferred to rely on the expertise of their doctor rather than on written information provided (Ducourneau 2005). The experts consulted at the Seville workshop also emphasised the fact that patients, as well as research volunteers, have varying needs relating to the quality and the quantity of information necessary to make their choice: some might require full and detailed information and would wish to consult different sources, whilst others might feel overloaded by so much information and would rely on their doctor. Moreover, patient information needs are likely to depend on the research or clinical use of a genetic test and on the characteristics of the condition or trait it identifies, since potential benefits and risks vary substantially. Finally, experts stressed that the roles of different professionals may vary according to the disease-specific service organisation and to the national or regional health care system.

*Initiatives to be taken*

Based on these considerations, during the Seville workshop, experts suggested that the question of how to realise the principle of free and informed consent in practice is still open.

They regret a lack of empirical evidence on the expectations of patients and/or donors with respect to pre-counselling, information and consent in clinical and research contexts. Clearly, empirical sociological and psychological research is required to better understand the needs of the different players, be it the patients, the research volunteers or medical personnel. Research efforts should take into consideration the context of utilisation of the tests.

Questions to investigate could be related to: the form and quality of information needed; the sources of information to be relied upon; the kind of pre-counselling expected; the role of doctors and other actors (e.g. psychologists); trust mechanisms, etc. Research on these questions should of course consider existing studies or reviews[6].

More interactive projects could also be envisaged to produce information on tests together with patients, or to establish procedures where information could be adapted to the needs of patients.

Parallel to these investigations, research on to what extent principles like informed consent are really guiding the practice of testing should be launched. As a matter of fact, the projects reviewed in our sample showed that some experts and professionals demand more quality in genetic counselling or fear that economic pressure on health care systems, together with the increased use of genetic testing, might endanger counselling. Are these fears realistic? On which aspects is counselling deficient and how could it be improved? These are questions that certainly deserve some attention.

*Players*

Social scientists, psychologists, Centres for human genetics

### 5.3.2. Generic consent for biobanks: a new concept in need of examination

*Background*

Biobanks – i.e. collections of biological samples (e.g. cells, tissue, blood, or DNA) and the related databases – are an important resource for medical research. But at the same time, they raise important ethical questions relating to data protection and consent. The use and development of biobanks is thus confronted with a dilemma: whereas biobanks offer important prospects for medical and pharmaceutical research, they have to protect the rights of those individuals whose samples and data are stored.

Several ethical committees and professional bodies have taken positions and developed recommendations, which should reconcile the apparently contradictory aims of the promotion of research and donor protection. For instance, the European independent expert groups states, in

[6] See for instance Corrigan 2003.

its 25 recommendations on genetic testing, that "the informed consent should be fully transparent with regard to the planned research, including policies on provision of test results to individuals and to the population, as well as to the handling of samples and the rights of sample donors" (European Commission 2004). In the same vein, the German National Ethics Council considers, in its position on biobanks, that "the collection of bodily substances from [the donor's] body and the gathering of personal data, in both cases for subsequent use in biobanks for the purposes of medical research, must be subject to the donor's consent. The consent is effective if the donor has the capacity to give consent, the consent is given voluntarily and the donor has been appropriately informed of the purposes, nature, significance and implications of the collection and use" (Nationaler Ethikrat 2004).

These recommendations do not exclude the use of genetic data for further research, but do recognize the importance of respecting the rights of donors. How can this principle be implemented? In practice, several forms of consent can be envisaged, ranging from specific consent for given research to presumed consent (consent is presumed in the absence of explicit refusal). According to some experts at the Seville workshop, the concept of generic consent (i.e. consent for the current research and for any further authorized research) might offer an intermediary and practical solution able to reconcile both the needs of research and the donors' rights. It might also offer a solution for the use of samples or data of deceased persons (without specific consent, samples and data of deceased persons cannot be reused for new investigations).

*Initiatives to be taken*

Recently, the British authorities acknowledged the concept of generic consent in the Human Tissue Act 2004 (see http://www.opsi.gov.uk/acts/acts2004/20040030.htm). The Act sets a baseline requirement for consent but does not require consent for the use of tissue in research to be project-specific. This decision is innovative and needs further regulation. Would such a solution be compatible with the legal frameworks of other European countries? There is clearly a need to do research on the implications of generic consent in the context of national legislations. Moreover, the introduction of such a concept would require reconsideration of the way in which information is delivered to patients or research volunteers, and more generally how counselling is conducted prior to the tests.

In a broader perspective, experts also called for the launching of participatory projects on the topic of generic consent for biobanking, involving both patients and the general public.

*Players*

Ethicists, ethic committees, law specialists

### 5.3.3. Persons judged incapable of giving consent: who should decide?

*Background*

The debate on who should decide for persons considered as lacking capacity to give consent (minors, mentally incapacited persons, people legally declared unable to give consent) is still open and subject to controversy. Some claim that free consent is an intangible principle, which should only be countered when it is clear that the person is unable to take a decision. According to this position, minors above a certain age limit or persons with a slight mental handicap should be asked for their consent. Others consider that the existing legislation should govern the situation: i.e. the responsibility for minors and for persons legally declared unable to give consent resides with their legal respondent and this also applies to medical decisions.

This question gives rise to a lively debate, involving medical personnel, families and, in some cases, the patients themselves. There are no fixed groups in opposition here, but varying group constellations according to the disease, the nature of incapacity to give consent (age or handicap), etc.

*Initiatives to be taken*

According to experts attending the Seville workshop, the question of persons lacking capacity to give consent requires further investigation, taking into account that no unique solution can be envisaged. Adjustments could be made according to the age of the patient (when the persons concerned are minors), the kind of diseases considered and the type of tests. This would imply, first of all, a categorisation of reasons for inability to give consent. Since the debate confronts diverging points of views – especially between professionals and family carers where professionals emphasise rights but families focus on responsibilities - interested parties and experts should also have the opportunity to present and discuss their positions in a constructive dialogue (stakeholder and public dialogues).

*Players*

Persons concerned (parents, patients, etc.), health and medical personnel, ethicists, jurists.

### 5.3.4. Values and norms: a reality-check

*Background*

Most projects considering the ethical and social aspects of genetic testing services are of a theoretical nature, discussing the implications of genetic tests alongside moral, societal and medical principles. The conclusions of these projects – whether research projects or participatory initiatives – consist mainly of statements, with general principles being expressed

such as “there should be no discrimination”, “patients should get professional counselling before and after the genetic test”, “data protection should be assured”, etc.

Experts who participated at the Seville workshop considered that more empirical research should be gathered in order to underpin (or not) these statements. To what extent and in what manner are these general principles respected in practice? For instance, does privacy mean that patients or research volunteers want to withhold their personal data? Under what conditions would they agree to reveal their data? Do patients and research volunteers wish to make their own decisions about undergoing a test or do they prefer to delegate this decision to doctors? How is the right to know (and not to know) considered by the persons concerned? What are their expectations about counselling? Do patients or volunteers have similar needs with regard to genetic testing services or do they have diverging expectations?

*Initiatives to be taken*

The experts at the Seville workshop shared the view that the many normative statements relating to genetic testing services should undergo a “reality-check”, based on empirical research. This reality-check would throw light upon how the general principles published in research documents or guidelines could be implemented in order to fit the expectations of the different players involved (patients, families, medical personnel and researchers). It would also provide an empirical basis for a fine-tuning between principles and practices, when necessary. As a first step, they suggested the review of existing evidence on specific topics. A second step would imply identification of the fields where a gap between principles and practice is felt and thus to launch surveys on patients’ or other players’ expectations with respect to genetic testing services.

Experts also acknowledged that the difficulty in implementing normative principles may have its origin in structural factors, such as the coexistence of a variety of regulatory mechanisms that can be contradictory, incoherent or poorly understood. They thus suggested that harmonization be sought and the implementation of regulatory frameworks by the concerned institutions and stakeholders be improved, where necessary.

*Players*

Sociologists, Socio-psychologists, public, health professionals

## 5.4. Genetic testing services as part of the health care sector

### 5.4.1. Studying the specificities and common factors of genetic testing services

*Background*

Genetic tests have been subject to many investigations about their ethical and social implications. The sample of projects selected by viWTA for the present study represents only a small amount of the published work. Few other medical techniques attract so much attention. However, many of the questions and issues relating to the ethical and social implications of genetic testing services are also subject to discussion in other medical domains. For instance, studies looking at counselling in the case of pre-implantation diagnosis highlight issues also relevant for other prenatal testing. The discriminatory potential of medicine is also discussed in other fields, such as HIV/Aids testing. And the drive for data protection, which is presented as a crucial issue of genetic testing services, is an essential guiding principle for any medical activity based on the Hippocratic oath.

Obviously, genetic tests are part of the health care system and, in this respect, share common features with other medical practices. At the same time, genetic testing services are far from being a standard practice. They open wide vistas for the diagnosis and treatment of certain diseases. But, on the other hand, genetic tests pose controversial and specific issues. These issues constitute a great challenge for the medical profession, as they question accepted medical principles and practices. Clearly, innovative answers have to be found in order to address these issues.

Some voices, however, criticize all the attention on genetic testing services, claiming that genetic tests concern a minority of diseases and that major diseases such as cancer also raise ethical and social questions, which are only marginally considered by professionals and authorities. As an answer to these critics, experts at the Seville workshop considered that the many efforts invested in the comprehension and recognition of the implications of genetic testing services for individuals, families and society should benefit not only this particular medical field and those affected by inherited diseases, but that it should reflect on and be integrated into other medical sectors where similar questions and issues may occur.

*Initiatives to be taken*

Experts present at the Seville workshop suggested that the specificities of genetic tests should be studied further, as should the common factors shared with other medical fields. This would permit the application of what has been learnt from other similar medical situations, but would still allow for identification of those areas that are unique to genetic testing and to apply appropriate action.

Experts also recommended launching research projects reflecting on the ways debates, research and recommendations addressing genetic testing services may benefit other health sectors. For instance, what can we learn from the whole discussion on free and informed consent for other medical tests or interventions? How far could the counselling set up in the domain of genetic testing services be transposed to other domains, or even merged in broader medical counselling? In order to address these questions, experimental projects of cross-disciplinary counselling could be set up and implemented. The idea would be to launch pilot projects where genetic counselling services would be merged with other types of medical counselling.

*Players*

Public health officers, medical personnel, geneticists, legal experts, policy makers, social scientists and ethicists, counsellors

### 5.4.2. Economic impact of genetic testing services

*Background*

Broad knowledge has been acquired on the technical and medical aspects of genetic testing services. There is, however, very little evidence on the economic impact of genetic tests for the health care system, although this is an important aspect for decision-making, along with others such as their effectiveness for fighting certain diseases.

Considering the general assumption that prevention is cheaper than cure, one could say that genetic tests may reduce health costs, or at least halt their increase. This, of course, is a very simplistic view. Genetic tests are related to specific diseases, which have their own particular patterns. Some diseases can be effectively avoided by preventive measures, but others require expensive treatments. And sometimes, cost-effective measures are in contradiction with ethical principles or, at least, put the concerned persons in fundamental ethical dilemmas (such as aborting after a prenatal positive test). Studying the costs – or savings – occasioned by genetic tests thus requires calculations to be made on a case-to-case basis, for each disease. Moreover, different kinds of genetic tests may have different economic consequences: diagnosis, predictive and screening tests have diverse economic consequences.

Considering the economic impacts of genetic testing services also implies an interdisciplinary approach, as costs are influenced not only by their clinical effectiveness and subsequent management, but also contain social and psychological dimensions. In fact, psychological reactions and behaviour of persons concerned when seeking genetic information may have a broad impact on the health care system and its costs. For instance, those found genetically at low risk of an illness may not participate in surveillance offered or may opt for behaviour which

increases the risk. On the other hand, those learning that they might (or will) develop a given disease may cause the health care providers to deal with a new sort of distress. These dimensions need to be taken into account when assessing the costs and benefits of genetic tests and related services.

Family implications of genetic testing also need to be considered when discussing their costs. Test results on an individual can give information on many other members of a family, so that economic impact should be assessed not only at the individual level but also at family level, and even in some cases at group level.

*Initiatives to be taken*

Experts attending the Seville workshop considered that a comprehensive image of the economic implications of genetic testing services is lacking, mainly because of the complexity of the issue and the difficulty of making sound comparisons. There is a clear need to obtain reliable information on the effectiveness of genetic testing services for different diseases, compared to other treatments or prevention strategies. According to the experts, knowledge should be sought on the economic impact of new treatments, which would not be available without genetic testing. In the same vein, investigation should be undertaken in order to find out how different kinds of tests (e.g. diagnostic, predictive or screening of populations) impact on health economics. For instance, will developments in screening technologies make treatment cheaper and easier, and should they thus be promoted? Or, on the contrary, should they be limited to certain population groups or certain diseases in order to control health costs?

Assessing the economic impacts of genetic tests should in any case have a broad focus, considering the health care system as a whole. This implies the assessment not only of the clinical effectiveness of genetic tests, but also the way social and psychological dimensions might influence the health care system in terms of delivery of services, human resources or possible side-effects on patients' and relatives' health.

*Players*

Health economics, Governments, health care providers, public health officers, pharmaceutical companies, insurance companies, ethicists, academia and scientific institutions, clinical and epidemiological research, health services research.

### 5.4.3. Population screening

*Background*

There are currently important pressures to expand population screening (neonatal, prenatal and carrier screening) as medical techniques are being developed. In some countries, formal rules

are in place and sets of criteria have been officially adopted, whereas in many others no formal procedures are in place and responsibilities have not been clearly allocated.

*Initiatives to be taken*

According to the experts present at the Seville workshop, each screening programme should be evaluated on a case-by-case basis before being implemented. Such an evaluation requires input from a broad range of evidence, which may thus imply that preliminary research and pilot projects have to be conducted. Monitoring is also recommended for ongoing programmes. The task of combining the evidence and perspectives is a challenging one and the use of criteria, although traditional in public health, can itself raise some difficulties and controversies. In this respect, it is suggested that participatory methods be implemented in order to define evaluation criteria[7].

Besides the issues around criteria and decision-making processes, other unresolved problems pertain to ethnicity, consent, the nature of acceptable benefits, the handling of incidental findings, etc. All these issues deserve further investigation, review and, more specifically, public participation.

*Players*

Health care specialists, ethicists, ethnologists, concerned groups.

### 5.4.4. Genetic tests on the Internet

*Background*

In its project-review phase, the viWTA project surveyed several public consultations on genetic testing services (mostly consensus conferences), where the citizens involved mentioned the risks related to the availability of genetic tests on the Internet without any control and counselling. In the same vein, an international survey made with University students as potential users of genetic tests showed that even though this population is rather enthusiastic about genetic tests, they would prefer their delivery through health services so as to ensure accessibility for all and counselling (Levitt 2001). Experts attending the Seville workshop shared the public preoccupations about free and uncontrolled availability of genetic tests on the Net, but were not inclined to be over-alarmist. They took a prudent position, recommending more investigation on the availability of genetic tests on the Internet, as well as on the views of the public on commercial genetic tests. They considered that the main challenge in this field is

[7] Within the AETMIS platform (see http://www.aetmis.gouv.qc.ca/en/), a project on criteria for population screening is currently under way. Criteria will be defined on the basis of both a review, focus groups and a debate within the APOGEE-Network.

finding ways to ensure oversight mechanisms in a field of activity which currently escapes both national and international efforts.

*Initiatives to be taken*

For the moment, there is no clear view on the actual practices relating to the sale of health genetic tests via the Internet[8]. Experts thus suggested that investigations be carried out – and regularly updated – on the kind of products available on the net and the people using them. They also called for a better understanding of their motivation to buy genetic tests. Moreover, studies should be undertaken in order to identify national and international rules able to minimise the risks of "e-genetic tests". International coordination efforts are also necessary, as the issue goes beyond national borders.

*Players*

Public health officers, geneticists, legal experts and ethicists

## 5.5. Genetic tests as a social phenomenon

### 5.5.1. Selection and enhancement: risk or opportunity?

*Background*

Genetic tests have been developed principally to identify individuals carrying genetic mutations leading to certain diseases, in order to offer to patients adequate treatment or prevention programs. However, as the number and accuracy of genetic tests increases, and as costs decrease, the potential to obtain genetic information about individuals for uses outside the medical sphere also increases. Thus, genetic tests could be used not only for strict medical purposes, but also for selection purposes, be it in medical or non-medical contexts.

The development and use of pre-implantation genetic diagnosis opened the discussion on the selection potentials of genetic testing services. Heated debates have taken place – and in some places are still on-going – on the legitimacy of selecting embryos on the basis of their genetic characteristics. Whereas a tendency can be observed across Europe towards the authorization of pre-implantation diagnosis in the case of severe hereditary diseases as an alternative to prenatal tests and abortion (negative selection), the discussion about the possibility of positive selection (i.e. the selection of an embryo with specific genetic characteristics) is still animated in

[8] Some reports have been published on the availability of commercial genetic tests (see for instance Human Genetic Commission 2003), but these have a broad scope and have been done on a national basis.

most European countries. In particular, the use of pre-implantation diagnosis by parents wishing to give birth to a baby to treat a sick sibling is harshly discussed in most European countries, especially since the British House of Lords declared lawful the creation of so-called "designer babies" or “saviour-siblings”, upholding an earlier court decision.

*Initiatives to be taken*

According to participants at the Seville workshop, the creation of designer babies through pre-implantation diagnosis merits further discussion and debate, involving geneticists, gynaecologists and the general public. There is an urgent need to undertake activities on this topic, as several European countries authorize – or will be soon authorizing – this practice (such as Spain, France, and Belgium). For instance, the public and concerned couples should be informed about the success rate of pre-implantation diagnosis, the efforts such a treatment implies, etc. Participatory projects should also be launched, in order to discuss crucial ethical and social questions related to pre-implantation diagnosis.

Other research trends and evolution in genetics demonstrate further risks of selection, not at birth but during a person’s lifetime. For instance, sport genomics offer promising prospects for a better understanding of the abilities of individuals for certain exercises or sports. Will athletes be selected not only on their results, but also on the basis of their genetic profile? Can we imagine other fields where individuals might be evaluated according to genetic characteristics, such as professional orientation? These questions might appear futuristic and not related to reality, but the experts at the Seville workshop recommend that our societies reflect on how to cope with non-medical genetic tests and the general principles to apply before these become reality.

*Players*

Ethicists, ethic committees, government, general public.

### 5.5.2. Discrimination: a reality?

*Background*

There has been wide discussion of genetic tests relating to the fears and risks of discrimination. For instance, fears have been expressed about employers or insurance firms requesting a genetic test to obtain information about a person’s genetic profile, in order perhaps to exclude a “bad risk”. As our knowledge of the human genome increases, there will certainly be further developments in the understanding of human genetics thus strengthening the fears of selection and discrimination.

Non-discrimination principles, which can be found in most guidelines and recommendations relating to genetic testing services, have been established as a response to these fears. For

instance, the EU Independent expert group stated that “data derived from genetic sources should not be used in ways that disadvantage or discriminate unfairly against individuals, families or groups in either clinical or non-clinical contexts, including employment, insurance, access to social integration, and opportunities for general well-being” (European Commission 2004).

*Initiatives to be taken*

Nevertheless, fears of discrimination are still present in many circles and among the general public. Experts at the Seville workshop considered that these fears should be taken seriously and that everything should be done to avoid any discrimination. However, they noted that the actual discussion occurs mainly at a theoretical level, without reference to concrete cases. The experts thus recommended that a reality-check be undertaken about possible discriminatory practices related to genetic testing. Can cases of discrimination be observed? How are non-discrimination principles enacted in the many guidelines and recommendation documents put into practice? What is the effectiveness of anti-discriminatory legislation? Such investigations would certainly be worthwhile in order to put the debate on discrimination on a factual basis.

*Players*

Social scientist, psychologists

### 5.5.3. Ethnicity

*Background*

Issues related to the use of ethnic information in health care are not unique to genetics, but are likely to be exacerbated in this field due to the variations in prevalence and distribution of mutation across populations. Ethnic information may thus be relevant for both clinical care and for screening purposes. Offering tests to high-risk groups represents gains in terms of efficiency, but may raise concerns in terms of equity in access to services, for example.

The debate on ethnicity has recently experienced a new turn, after the decision of the European Patent Office to grant a patent on the BRCA2 gene test when used on Ashkenazi Jewish women. Delivering patents restricted to certain groups of population raises questions in terms of accessibility: whereas patients who do not belong to the ethnic group included in the patent can be entitled to a free test, those who belong to the ethnic group will only be granted a genetic test if their health provider pays for a licence.

*Initiatives to be taken*

The experts at the Seville workshop considered that population based genetic testing and pharmacogenetics must be thoroughly studied and debated, in terms of both equity and

efficiency. Implementing screenings only in certain population groups may increase inequalities, as those considered at risk may benefit from preventive measures whilst others are excluded from prevention programs. These screening programs also raise some efficiency- related questions: are the efforts and resources invested compensated by a reduction of risks and/or an early cure, avoiding more costly and invasive treatment? The same kind of questions arises when drugs are developed for specific ethnic groups: should we fear inequalities in the availability of treatments?

The question of ethnicity with respect to patenting and marketing also deserves further scrutiny. Experts call above all for a public dialogue, which should include pharmaceutical companies.

*Players*

Social scientists, medical professionals, healthcare specialists, Pharmaceutical companies; general public.

## 5.6. Building on research initiatives

### 5.6.1. A structured overview of European Union funded research

*Background*

European Research has subsidised major and innovative research related to genetic testing services and their ethical and social implications in its different Framework programmes. Whereas some projects consider genetic testing services as a whole (this is the case for instance of the EUROGENTEST Network of Excellence), others focus on specific tests or diseases (as for example the project “Prenatal testing for Huntington's disease” in FP 4), on the ethical and social implications of genetic testing (e.g. the EUROSCREEN project in FP 4; part of the viWTA sample). Still others consider genetic testing services within a broader discussion on biotechnology or bioethics (see for example the COB - Challenges of Biomedicine: Socio-cultural contexts, European governance and bioethics – project in FP 6).

All these projects have produced very valuable results. However, a comprehensive and structured overview of their results is lacking, each project carrying its own rationale and having its own “career”.

*Initiatives to be taken*

The experts present at the Seville workshop share the view that the European funded projects have produced valuable results, which have to be considered when discussing the ethical and

social implications of genetic testing services. They thus suggested that a synthesis report be launched, aiming at providing an inventory of all European funded research projects addressing in one form or another the ethical and social implications of genetic testing services. This inventory would of course systematise the issues considered, as well as the results and recommendations of the projects. Based on this inventory, the European Commission could draw conclusions for further research or for the definition of a common strategy on genetic testing in Europe. Based on this inventory, the EU may also adapt recommendations, decide on an implementation plan and check the results of agreed action.

*Players*

European Commission, ethical and social experts

### 5.6.2. Bringing EUROGENTEST to the people

*Background*

As a network joining the major players in Europe in the field of genetic testing services and working on the improvement of genetic testing services throughout Europe, EUROGENTEST might offer a productive and necessary platform for a discussion between the public, geneticists and researchers. During the Seville workshop, experts thus suggested the launching of participatory projects on what is going on in the EUROGENTEST Network of Excellence.

*Initiatives to be taken*

Launching participatory projects involving the general public might be an occasion to examine norms and assumptions relating to genetic testing services, an issue covered by Units 4 and 6 of EUROGENTEST.

*Players*

Experts and researchers of the EUROGENTEST Network of Excellence, TA community

## 6. Literature


Borry Pascal, Fryns JP, Dierickx Kris, 2005. Predictive genetic testing in children. In: de Bouvet A (ed.), *Ethical issues in predictive medicine.* Paris: éditions John Libbey.

Cambon-Thomsen, Anne, 2003. Assessing the impact of biobanks. *Nature Genetics (Correspondence),* 34, (1) : 25-26

Cambon-Thomsen, Anne, 2004. The social and ethical issues of post-genomic human biobanks*, Nature Reviews Genetics*, 5: 866-873

Chadwick, Ruth et al., 1997. *EUROSCREEN Final report. Genetic Screening: Ethical and Philosophical Perspectives,* Euroscreen, February 1997).

Chadwick Ruth et al. 1999. *Genetic screening and testing: toward community policy on insurance, commercialisation and promotion public awareness (Euroscreen 2),* Final report.

Corrigan, Oonagh, 2003. Empty ethics: the problem with informed consent, *Sociology of Health & Illness,* vol. 25, No. 3, pp. 768-792.

Decker, Michael and Miltos Ladikas (eds.), 2004. *Bridges between Science, Society and Policy. Technology Assessment – Methods and Impacts,* Berlin: Springer Verlag.

Doucournau, Pascal, 2004, Le consentement à la recherche en génétique humaine : entre dispositif justifié et conflits de valeurs. In F. Bouchayer, G. Cresson, S. Pennec, F.X. Schweyer (eds), *Normes et valeurs dans le champ de la santé*, Paris: Editions de l'Ecole Nationale de Santé Publique, pp.273-280

Doucournau, Pascal, 2005, Le consentement à la recherche en épidémiologie : un « rituel de la confiance » en question, *Sciences Sociales et Santé,* vol. 23. n°1. 2005. pp 5-36.

EUROGENTEST, 2003. Genetic testing in Europe: Network for test development harmonisation, validation and standardisation of services, KU Leuven, Belgium.

European Commission, 2004. *25 Recommendations on the ethical, legal and social implications of genetic testing services,* Brussels: EC Directorate General for Research.

Grunwald, Armin (ed), 2002. *Technikfolgenabschätzung. Eine Einführung.* Edition Sigma, Berlin.

Harris, Rodney (ed.), 1997. Genetic Services in Europe. A comparative study of 31 countries by the Concerted Action on Genetic Services in Europe (CAGSE), *European Journal of Human Genetics,* 5 (suppl 2): 1-220, 1997

Hennen, Leonard et al. 2000. *Genetic diagnostic - status and prospects. Status report,* TAB Working report, No 66, Berlin.

Hennen Leonard and Arnold Sauter, 2004. *Preimplantation diagnostics: Practice and statutory regulation in seven selected countries. Status report,* TAB Working report, No 94, Berlin.

Hirtzlini, Isabelle et al., 2003. An empirical survey on biobanking of human genetic material and data in six EU countries. *European Journal Human Genetics*, 2003, 11 (6):475-488,

Human Genetics Commission, 2003. Genes direct: Ensuring the effective oversight of genetic tests supplied directly to the public. A report by the UK Human Genetic Commission, March 2003.

Jonas, Susanna et al. 2002. *Prädiktive Humangenetische Diagnostik bei hereditärem Mamma- und Kolorektalkarzinom.* Wien: Institut für Technikfolgenabschätzung.

Joss, Simon and John Durant (eds.), 1995. *Public Participation in Science: The Role of Consensus Conference in Europe.* London: Science Museum.

Levitt, Mairi, 2000. The Gene Shop at Manchester Airport, *New Genetics and Society,* vol. 19 no. 2, pp. 77-87.

Levitt, Mairi, 2001. Let the consumer decide? The regulation of commercial genetic testing. *The Journal of Medical Ethics,* 27:398-403.

Nationaler Ethikrat, 2004. *Biobanks for Research: Opinion,* Berlin.

Nuffield Council on Bioethics, 2003. Pharmacogenetics: ethical issues.

Rathenau Institute, 1995. *Predictive genetic research, where are we going?,* Rathenau Report.

Rippe, Klaus Peter et al., 2004. Pharmakogenetik und Pharmakogenomik, TA-SWISS Report 48/2004, Berne

## Appendix I: Participants of the project

### Participants framing workshop, 2 March 2005

**Bjorn Bedsted,** Danish Board of Technology, Denmark

**Prof Jean-Jacques Cassiman,** Center for Human Genetics, KU Leuven, Belgium

**Domenico Coviello,** Laboratory of Medical Genetics, Milan, Italy

**Prof Kris Dierickx,** Centre for Biomedical Ethics and Law, KU Leuven, Belgium

**Dolores Ibarreta,** IPTS, Spain

**Koos Van der Bruggen,** Rathenau Institute, The Netherlands

**Tinne Vandensande,** Koning Boudewijn Stichting, Belgium

### Participants Seville Workshop, 26-27 May 2005

**Sergio Bellucci,** Centre for Technology Assessment, Switzerland

**Ingeborg Blancquaert,** Agence d'évaluation des technologies et des modes d'intervention en santé (AETMIS), Canada

**Pascal Borry,** Centre for biomedical ethics and law, Faculty of Medicine, Leuven University, Belgium

**Angela Brand,** German Center for Public Health Genetics (DZPHG), MPH, Germany

**Anne Cambon-Thomsen,** Inserm U 558: Epidemiology and analyses in public health, Faculté de Médecine, Toulouse, France

**Dirk Carrez,** European Association for Bio-Industries, Brussels, Belgium

**Prof. Kris Dierickx,** Centre for biomedical ethics and law, Faculty of Medicine, Leuven University, Belgium

**Dolores Ibarreta,** DG JRC, Institute for Prospective Technological Studies, Spain

**Susanna Jonas,** Austrian Academy of Sciences, Institute of Technology Assessment, Austria

**Alastair Kent,** Genetic Interest Group, Unit 4D, London, United Kingdom

**Lars Klüver,** The Danish Board of Technology, Denmark

**Mairi Levitt,** Cesagen, Furness College, UK

**Prof. Alexandre Mauron,** Bioethics Institute, Faculty of Medicine, University of Geneva, Switzerland

**Helen Statham,** Centre for Family Research, University of Cambridge, United Kingdom

**Koos Van der Bruggen,** Rathenau Instituut, The Netherlands

**Robyns Wauthier,** Assuralia, Belgium

## Questionnaire's respondents

**Bjorn Bested,** Danish Board of Technology, Denmark

**Ingeborg Blancquaert**, Agence d'évaluation des technologies et des modes d'intervention en santé (AETMIS), Canada

**Anne Cambon Thomson,** Inserm U 558: Epidemiology and analyses in public health, Faculté de Médecine, Toulouse, France

**Carlo Castellani,** Centro Fibrosi Cistica, Ospedale Civile Maggiore, Verona, Italy

**Ruth Chadwick,** University of Central Lancashire, Centre for Professional Ethics, United Kingdom

**Prof. Kris Dierickx,** Centre for Biomedical Ethics and Law, KU Leuven, Belgium

**Gerry Evers-Kiebooms**, Psychosocial Genetics Unit and Clinical Genetics Unit, University Hospital KU Leuven, Belgium

**Leonard Hennen,** Büro für Technikfolgen-Abschätzung beim Deutschen Bundestag TAB, Germany

**Prof. Alexandre Mauron,** Bioethics Institute, Faculty of Medicine, University of Geneva, Switzerland

**Jörg Naumann,** The Deutsches Hygiene-Museum Dresden, Germany

**Adrian Rüegsegger,** Centre for Technology Assessment TA SWISS, Switzerland

**Helen Statham,** Centre for Family Research, University of Cambridge, United Kingdom

**Tinne Vandensande,** Foundation King Baudoin, Belgium

**Koos Van der Bruggen**, Rathenau Instituut, The Netherlands

**Hub Zwart,** University of Nijmegen, Centre for Society and Genomics, The Netherlands

## Project staff

**Danielle Bütschi Häberlin,** senior consultant (report)

**Peter Graller,** viWTA (communications co-ordinator)

**Mark Hongenaert,** Timeout (moderation)

**Stef Steyaert,** viWTA (project manager)

**Els Van den Cruyce,** viWTA (project assistant)

**Willy Weyns,** viWTA (project manager)

## Appendix II: Defining the project: the "framing workshop" (Brussels, 2nd March 2005)

As a first step of the project, viWTA organised a "framing workshop". Eight experts, members of the TA community or of the EUROGENTEST Network of Excellence, were invited to Brussels. Their task was to prepare the Seville international expert workshop, which would deliver the main input for the project and the report.

The first point of discussion concerned the drafting of the topic. First, it was decided to look at TA and other similar projects", that is, according to our definition of TA, projects which have an advisory aim (for stakeholders or policy-makers) and which use either scientific, interactive or communication methods. Second, participants decided that genetic testing services should be broadly considered, i.e. the project should look at scientific aspects of genetic testing (i.e. genetic tests developments), the testing itself (as a technical / medical intervention) and the services before, during and after testing (e.g. genetic counselling).

The framing workshop's participants also defined the questions to be addressed within the project. Based on these discussions, the following research questions were defined:

- Overview of the positions in Europe towards the social and ethical aspects of genetic testing services
- The arguments used
- Information about which groups / players take what position
- Identification of gaps, with a view towards further research or activities

Second, the framing workshop participants decided that the discussion and interaction during the expert workshop should be based on a review of projects, in which the following information would be made available:

- The type of genetic services considered by the project (category of genetic tests, time of testing, field of application, initiative for the test)
- Methodology of the study (scientific, interactive or communication project)
- Issues covered in the project and, for each issue, the existing positions, the arguments used and the players involved.
- Impacts of the project

This information would be gathered through a questionnaire sent by viWTA to the selected projects before the Seville International Expert Workshop.

Finally, the framing workshop participants briefly addressed the form of the Expert workshop planned by viWTA in its project design. They suggested that the workshop should aim at discussing and complementing the results of the questionnaire. It should conclude by elaborating recommendations to be distributed to decision-makers. They consequently

suggested that project managers, who could refer to their own experience and results, be invited, as well as scientists and interest group representatives who hold a comprehensive view of the topic and whose role would be to broaden the discussions. The framing workshop participants stressed the importance of inviting a broadly based expert panel and not to restrict it to the Technology Assessment community.

## Appendix III: Preliminary questionnaire for reporting about projects on ethical and societal aspects of genetic testing services

**1. General information about the project**

| **1.1. Title of the project:** |
|---|

| **1.2. Institution / Organisation in charge of the project:** |
|---|

| **1.3. Participants / members of the project (please, specify for each participant/member its function in the project):** |
|---|

| **1.4. Abstract of the project** *(if necessary, use a separate page)*: |
|---|

**2. Specification of genetic testing services considered in the project**

| **2.1. What type of genetic tests were involved in the project?**<br>*Indicate the categories that were considered, give comments when necessary* | |
|---|---|
| Category | |
| O | Diagnostic test: clinical diagnosis |
| O | Diagnostic test: defining diseases |
| O | Predictive test: late onset disease |
| O | Predictive test: predisposition |
| O | Pharmacogenetics |
| O | Nutrigenomics |
| O | Screening of populations |
| O | Other *(specify):* |
| Comments *(if necessary, use a separate page)*: | |

| **2.2. Moment of testing: Was the project focussing on specific moments of genetic testing?**<br>*Indicate the categories that were considered, give comments when necessary* | |
|---|---|
| Category | |
| O | Pre-implantation |
| O | Pre-natal |
| O | Neonatal |
| O | Minors |
| O | Adults |
| O | Other *(specify):* |
| Comments *(if necessary, use a separate page)*: | |

| **2.3. Field of application: What kind of applications were implied?**<br>*Indicate the categories that were considered, give comments when necessary* | |
|---|---|
| Category | |
| O | Health |
| O | Quality of life (sports, wellness, …) |
| O | Forensic (related to crime) |
| O | Paternity |
| O | Family Planning |
| O | Other *(specify)*: |
| Comments *(if necessary, use a separate page)*: | |

| **2.4. Testing initiative: Which parties took initiative for genetic testing in the project under consideration?**<br>*Indicate the categories that were considered, give comments when necessary* | |
|---|---|
| Category | |
| O | Doctors, medical personnel |
| O | Individual initiative |
| O | Public authorities |
| O | Employer |
| O | Insurance company |
| O | Commercial organisation |
| O | Other *(specify):* |
| Comments *(if necessary, use a separate page):* | |

**3. Methodology and approach of the project under review**

| **3.1. Methodology of the project: which methodologies were used:**<br>*Indicate the methodologies that were used, give comments when necessary* | |
|---|---|
| Category | |
| O | Literature analysis |
| O | Trends extrapolation, modelling, etc. |
| O | Expert workshop, delphi, etc. |
| O | Survey |
| O | Participatory method |
| O | Other *(specify): expert interviews* |
| Comments *(if necessary, use a separate page)*: | |

| **3.2. If the project was participatory, which groups of people were involved?**<br>*Indicate the categories that were involved, give comments when necessary* | | |
|---|---|---|
| Category | | Number involved |
| O | Citizens (public) | |
| O | Patient groups | |
| O | Experts | |
| O | Stakeholders *(specify):* | |
| O | Other *(specify):* | |
| Comments *(if necessary, use a separate page)*: | | |

**4. Aspects / issues that are covered in the project under review**

**4.1. Which aspects / issues are considered in the project? At which level of analysis have they been considered: individuals, community (family, enterprise, etc.) or society?**

*Indicate the aspects / issues under consideration and the level of analysis. Give comments when necessary.*

| | Aspect / issue | Level of analysis (Society, community and/or individuals) |
|---|---|---|
| O | 1. Right to know (or not to know) | |
| O | 2. Free and informed consent | |
| O | 3. Privacy | |
| O | 4. Ownership and control over results and genetic material | |
| O | 5. Discrimination (gender, ethnicity, …) | |
| O | 6. Accessibility to genetic tests | |
| O | 7. Geneticisation of society | |
| O | 8. Commercialisation of genetic testing services | |
| O | 9. Costs to health sector | |
| O | 10. Quality of genetic testing services.<br>(Who should do the testing and counselling?) | |
| O | 11. Religion | |
| O | Other *(specify):* | |

Comments or precisions about the aspects/issues considered in the project *(if necessary, use a separate page)*:

**4.2. For each of the aspect / issue considered by the project under review, which points of view have been revealed (consensus, dissensus, in evolution), which groups take what position, which arguments are used ?**

*Specify the aspects / issues under consideration, the points of view, the groups involved and their position, and the arguments used. If necessary, use a separate page*

| Aspect / issue under consideration | Points of view (consensus, dissensus, in evolution) | Which group take what position | Arguments used |
|---|---|---|---|
| | | | |
| | | | |
| | | | |
| | | | |
| | | | |
| | | | |

**5. Impacts of the project, contribution of the project to the debate on the ethical and societal implications of genetic testing services**

**5.1. What impact did the project have, how did it contribute to the development of the ideas or practices in the field of genetic testing services?**

*Indicate the impacts that occurred, and explain*

| Type of impact | | Explanation |
|---|---|---|
| O | Enhancing public awareness by media coverage | |
| O | Enhancing insight and attitudes of patients | |
| O | Stimulating the dialogue between society and science, between society and health professionals | |
| O | Mediation between antagonists, diminishing polarisation | |
| O | Enhancing the relationship between private and public sector R&D | |
| O | Developing guidelines, best practices | |
| O | Putting the issue on the political agenda | |
| O | Initiating / stimulating change in legislation | |
| O | Contributing to other (similar) discussions related to health & medicine | |
| O | Other *(specify):* | |

**6. Other points/comments you would like to make about the project?**

**Thank you very much for your participation**

Please return the questionnaire **before 18 April 2005** to

viWTA
Vlaams Parlement
1011 Brussel
België
Willy.weyns@vlaamsparlement.be
Stef.steyaert@vlaamsparlement.be
Els.vandencruyce@vlaamsparlement.be

## Appendix IV: The International Expert Workshop, Seville, 26-27 May 2005

On 26 and 27 May, the Flemish Institute for Scientific and Technological Assessment (viWTA) and the Institute for Prospective Technological Studies (IPTS) invited 16 international experts to Seville to take part in a workshop dedicated to the discussion and evaluation of research efforts relating to the ethical, social and governance aspects of genetic testing services. The aim of the workshop was to draw lessons from projects that address the implications of genetic testing services and to find out: which issues are covered by these projects? What positions on these issues do they reveal? Which issues need new or further action in order to support decision-making processes in research, clinical practices, health care delivery or State regulation?

A sample of 18 projects, selected by viWTA for their variety in terms of methodology, type of genetic tests and geographical scope, provided core material for the discussion of these questions at the Seville workshop. Before the workshop, the experts received a rough description of these projects and their results. Based on this overview, they discussed, in a first round, the social, ethical and governance issues of genetic testing services. The aim here was not to ascertain their point of view on these issues, but to discover which issues have already been considered in the many research projects and dialogue initiatives addressing genetic testing services, as well as to get a picture of the positions and points of views of different key-players on these issues.

Based on these assessments and discussions, the experts then identified, in a second round, issues for which further or new initiatives (research or participatory projects) should be taken since there might be conflicting positions on a given issue, there has not been enough investigation, or it is a new issue. This second part was the crucial element of the workshop, as it was meant to define the main outcome of the report. It must be remembered that the whole project is intended to indicate new avenues of research and action to the EUROGENTEST Network of Excellence and to decision-makers involved in the research, provision and regulation surrounding genetic tests.