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REVIEW

Provision of genetic services in Europe: current practices and issues

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This paper examines the professional and scientific views on the social, ethical and legal issues that impact on the provision of genetic services in Europe. Many aspects have been considered, such as the definition and the aims of genetic services, their organization, the quality assessment, public education, as well as the partnership with patients support groups and the multicultural aspects. The methods was primarily the analysis of professional guidelines, legal frameworks and other documents related to the organization of genetic services, mainly from Europe, but also from USA and international organizations. Then, the method was to examine the background data emerging from an updated report produced by the Concerted Action on Genetic Services in Europe, as well as the issues debated by 43 experts from 17 European countries invited to an international workshop organized by the European Society of Human Genetics Public and Professional Policy Committee in Helsinki, Finland, 8 and 9 September 2000. Some conclusions were identified from the ESHG workshop to arrive at outlines for optimal genetic services. Participants were concerned about equal accessibility and effectiveness of clinical genetic services, quality assessment of services, professional education, multidisciplinarity and division of tasks as well as networking. Within European countries, adherance to the organizational principles of prioritization, regionalization and integration into related health services would maximize equal accessibility and effectiveness of genetic actions. There is a need for harmonization of the rules involved in financial coverage of DNA tests in order to make these available to all Europeans. Clear guidelines for the best practice will ensure that the provision of genetic services develops in a way that is beneficial to its customers, be they health professionals or the public, especially since the coordination of clinical, laboratory and research perspectives within a single organizational structure permits a degree of coherence not often found in other specialties.
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Introduction

The rapid development of our understanding of molecular genetics has created new possibilities to diagnose genetic disorders. It has also made presymptomatic, carrier and prenatal testing of family members possible in a growing number of disorders. Predictive testing for late onset diseases may be considered upon request of the person at risk. Even more, the rapid development of our understanding of molecular genetics has permitted a better understanding of the pathogenesis of a growing number of diseases and has changed the way in which we think about health and illness. This has increased public awareness of

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hereditary diseases and consequently the expectations regarding genetic prediction and therapy of genetic diseases. There are an increasing number of families requesting genetic services and more physicians prescribing genetic tests, usually highly specialized and expensive investigations. Simultaneously, there are demands in many European countries to cut down the costs of the public health care. All this has created a situation where professional guidelines for the provision of genetic services are needed.

In 1997, the provision of genetic services in the European countries was evaluated by the Concerted Action on Genetic Services in Europe (CAGSE). This evaluation revealed that the practices, resources as well as traditions in the different countries varied considerably. The need for guidelines was clearly stated in the CAGSE report: 'collaborators recognized the need for internationally agreed and published principles to help national and regional health departments plan genetic services to act as a service, research and educational resource in each country'.[1]

The same year, the European Society of Human Genetics (ESHG) nominated a Public and Professional Policy Committee (PPPC) for developing the ESHG's policies on social, ethical, and legal issues of human genetics and the relationships between professional geneticists and the public. The Committee decided to start its work by formulating, professional guidelines in the field of human genetics concerning different topics, one of which was guidelines for the provision of genetic services in Europe.

The present document aims to examine the professional and scientific views on the social, ethical, and legal issues that impact on the provision of genetic services in Europe. For this purpose, many aspects have been considered, such as the definition and the aims of genetic services, their organization, the quality assessment, public education, as well as the partnership with patients support groups, and the multicultural aspects.

Methods

The method used for analyzing the professional and scientific views on the social, ethical, and legal issues that impact the provision of genetic services was primarily the collection and analysis of existing professional guidelines, legal frameworks and other documents related to the organization of genetic services, mainly from Europe, but also from USA and international organizations. Then, with the help of the existing guidelines and a review of literature, the method was to examine the background data emerging from an updated report produced by the Concerted Action on Genetic Services in Europe (CAGSE), as well as the results of discussions held during an international workshop. This workshop was organized by the European Society of Human Genetics Public and Professional Policy Committee in Helsinki, Finland, September 8 and 9, 2000. The purpose of the workshop was to identify, from a professional viewpoint, the most important/pressing/burning ethical issues relating to the provision of genetic services in Europe. The formal workshop presentations covered the following themes: the aims and scope of clinical genetic services, organization and human resources, quality assessment of clinical genetic services, and challenges of present and future clinical genetic services. Small multidisciplinary groups were convened to take these discussions further. Their initial task was to explore the genetic services needs in the countries represented and to consider the extent to which these needs were currently being met. Following the small group sessions, conclusions were fed back to the whole group where there were opportunities for further discussion.

A group of 43 experts from 17 European countries was invited. These experts were representatives of the seven following sectors:

(1) Medical Genetics
(2) Human Genetics Societies
(3) Ethical, Legal, and Social Issues
(4) Support Groups
(5) Biotechnology/Pharmaceutics
(6) Insurance/Employment
(7) European Union Institutions

A first background document was discussed during the workshop. A second document, including discussions of the workshop, was sent for comments to representatives of the human genetic societies and European experts in the field of clinical genetic services, as well as to all ESHG members. This document was also put on the ESHG website (www.eshg.org) for public consultation and discussion. The final document was approved by the ESHG board.

National and international regulatory frameworks

To date, within different European countries, regulations on the provision of genetic services are differently organized (see Appendix A). The variety of regulations seems to be due to the fact that genetic testing is often considered directly related to health-care services. However, in a comparative study of 31 countries by the Concerted action on genetic services in Europe, Harris and Reid[2] have reported that in European countries where genetics is well established, a legal framework exists within which the services operate. The main recommendations made by the members of the concerted action were included in three principles:

- Official recognition of the specialty of medical genetics at a national level, and national strategic planning by medical genetics organizations linked to other specialties, to patient support organizations, and to government.

- Development of regional medical genetic centers as an important point of delivery of specialist genetic care in collaboration with other specialties, community and other medical services.
- Joint education and training programs to promote the teaching of genetics to medical and other students, and training programs with assessment for specialist geneticists and for other health-care workers.

More recently, the European Society of Human Genetics (2001) recommended a *Formal recognition of medical genetics as a medical specialty in Europe* in order 'to aid the provision and development of genetic services for individuals and families in Europe'. WHO[2] also recommended different measures for developing and strengthening medical genetics services, as well as to assist member states in establishing educational programs for the teaching of medical genetics.

A number of international bodies have published recommendations about the rights of individuals as they relate to developments in genetics. Examples include the Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine by the Council of Europe;[4] the Universal Declaration of the Human Genome and Human Rights by the United Nations Educational, Scientific and Cultural Organization (UNESCO);[5] the Proposed International Guidelines on Ethical Issues in Medical Genetics and the Provision of Genetic Services by the World Health Organization (WHO);[6] the World Medical Association Declarations (1992, 1995, 2000);[88–90] OECD[7] and the Charter of Fundamental Rights of the European Union (2000). All these texts stipulate that the medical application of genetic knowledge must be carried out with due regard to the general principles of medical ethics: doing good to individuals and families, not doing harm, offering autonomy of choice after information is given, and facilitating personal and social justice.

Issues

Definition and aims of genetic services

'Identifying indicators of quality, effectiveness or success of genetic services requires definition of the overall aims of clinical genetic services and what constitutes the services'.[8]

Even though human genetics in many European countries in the first half of the 20th century concentrated to eugenic aims, the goal has since then been to serve the interest of the families with an inherited disorder as clearly phrased by Fraser.[9] In the 1980 s, the general objective of genetic services was more precisely defined 'to help people with a genetic disadvantage to live and reproduce as normally and as responsibly as possible'.[10] Nowadays, the aim of a genetic service is often seen as to respond to the needs of individuals and families, particularly their wish to know whether or not they are at risk of developing a genetic disorder or of bearing an affected child. A primary responsibility in genetic counseling is to provide information as accurate as possible on diagnosis and chance of recurrence within the family. The tradition in genetic counseling is to be nondirective. However, it appears also important to maintain a proper balance between professional 'duty of care' and personal 'autonomy', that can include choosing to leave the decision-making to a professional team that the patient trusts.

The Clinical Genetics Committee of the Royal College of Physicians of London[11] defined three objectives of a clinical genetic service: (1) For persons who are affected, or who are referred because of a genetic risk, to make the genetic diagnoses, pedigree analyses and estimates of risk of transmission. These are necessary for genetic counseling and to guide preventive and therapeutic actions; (2) To support the identification and surveillance of relatives who are at risk for serious genetic disorders, but who may not have been directly referred, so that they may receive well-informed genetic counseling and guidance on preventive and therapeutic actions if required; and (3) to provide support to family members, both to those affected and unaffected. The British Clinical Genetics Society (2000) documented in detail the responsibilities of a clinical geneticist. Particular emphasis was placed on follow-up, support, coordination of health surveillance, and services to extended families. As for the Council of Regional Networks for Genetics Services (USA),[12] it distinguished three types of genetic services: (1) family-focused services; (2) population-based services; (3) clinical laboratory services.

Clinical genetics has emerged as a specialty in medicine. In fact, a genetic service is a specialized service provided in tertiary centers, accessed by self-referral or referral from consultant physicians and others including general practitioners, for patients and relatives with complex or rare conditions, and serving a wide geographic area. A genetic service is distinguished by the fact that diagnosis, investigations, counseling, and support is given for disorders affecting any organ system or at any age and records are sometimes kindred based and multigenerational, which requires extra-care for data protection. This imposes unique disciplines and requirements on the molecular diagnostic laboratory, which distinguishes it from other categories of clinical laboratory. The family is the unit of study in contrast to the individual. This will remain true even when mutation detection entirely supersedes linkage analysis.[13] Furthermore, inheritance across generations and in the extended kindred gives the information generated by the genetic laboratory a lasting relevance. It places on a laboratory a responsibility for long-term and careful storage and retrieval of clinical information.[13]

Finally, genetic services comprise multidisciplinary groups of medical and nonmedical disciplines such as in clinical setting geneticists, psychologists, genetic

counselors, genetic nurses and in laboratory setting biologists, bio-statisticians, specialized technicians, and administrative staff.

Services provided by clinical geneticists Clinical genetic services generate activities of varying complexity which range from single consultations for diagnosis and genetic counseling to clinical and laboratory investigations of other family members. Clinical genetic services also carry out pre- and postnatal diagnosis and carrier testing, bank DNA and other biological samples, accept inquiries, are committed to education and support community-based services.

Diagnostic consultations Traditionally genetic services have focused on single gene disorders, chromosomal abnormalities, malformation, and mental retardation syndromes and infertility problems including the provision of laboratory investigations. Much of this work has been linked to obstetric and pediatric practice and to the management of an individually rare but collectively significant group of genetic disorders. In recent years, these services have taken on an increasing workload. The new molecular genetic techniques have simplified some parts of the work (mainly diagnostic) and simultaneously have created new possibilities and new needs. This is particularly true in the area of predictive testing for neurogenetic diseases and hereditary cancers. In fact, even though familial cancers may be a small proportion of the total of cancers, they represent a considerable number by comparison with the rare disorders traditionally seen by clinical geneticists. Many families are now referred because of the possibility that they might fall into a hereditary subset of a common disorder.

As there are of thousands of syndromes and diseases, and as several new ones are described all the time in the medical literature, the only way to offer accurate services is to have experienced clinical geneticists and syndromologists, and to know how to better use specialized databases which help making more clinical diagnoses. Some of them are free of charge on Internet like OMIM (http://www3.ncbi.nlm.nih.gov/Omim/), Geneclinics (http//geneclinics.org/) or Orphanet (http://www.orpha.net). Others are distributed on CD-ROM like LDDB (http://www.hgmp.mrc.ac.uk/DHMHD/lddb.html) or POSSUM (http://www.possum.net.au/). In many countries, public Internet databases in native language are available.

It has been recommended that physicians and other health-care professionals in other clinics caring for these patients should have an easy access to diagnostic consultations.[2,14] This could optimally happen so that the clinical geneticist comes to see the patient/family, gives diagnostic suggestions and helps the physician to find recent and reliable information on the disease. Whether or not the diagnosis can, according to this advice, be ascertained, the family should be offered the possibility of genetic counseling.

If the accessibility to a genetic service is not available, another option that is proposed is the organization of consultations through regular or electronic mails: the physician may send data, photographs of the diagnostically problematic case to a clinical genetic unit in his/her own country or elsewhere.[15] In the future, expert centers may offer teleconsultation for a quick opinion or in case of a long-distance consultation. Teleconsultation may also be used for a second opinion between two professionals. It has been suggested that answering such inquiries should be an accepted part of the work load of clinical genetic units, ample time should be appointed to this work and this work should be considered for staffing and funding.[16]

Information and counseling The provision of genetic counseling is a defining characteristic of clinical genetic services. Genetic counseling has been defined as a communication process, which deals with the human and psychological problems associated with the occurrence, or risk of occurrence, of a genetic disorder in the family.[9] This process involves an attempt by one or more appropriately trained persons to help the individual or the family to (1) understand the medical facts, including the diagnosis, the probable course of the disorder and the available management; (2) appreciate how heredity contributes to the disorder and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.[9] This definition covers all types of genetic counseling,[17] as well as the counseling offered in the context of a multidisciplinary approach to predictive testing involving professionals from psychosocial disciplines.[18]

Many policy statements state that genetic counseling should be available to all, in particular to families with rare hereditary diseases as well as their close relatives. Families with common hereditary diseases, common malformations, and multifactorial diseases should also be offered genetic counseling services or counseled by other medical specialists. Some authors consider that much of the counseling of common problems like increased risk of chromosomal anomalies or preliminary evaluation of the possibility of hereditary cancer in a family, could be performed by specifically trained non-MD health-care providers or nongenetic specialist MDs outside genetic centers.[16]

It is generally agreed that families should decide themselves whether they want to be counseled or not and have free choice on where to go for the genetic

counseling (eg, some families may not want to discuss these issues in the same hospital where they are treated for the disease).[19] Traditionally, confidentiality has been and should continue to be one of the principles on which the relationship between patients and physicians is built. However, the arrival of medical genetics is likely to affect the way in which this principle is interpreted and how it operates within best clinical practice. Medical genetics is often concerned with patients in relation to their families and counseled patients may be asked to share the result of their genetic test with family members. It has been recommended that physicians should endeavor to explain to patients that sharing this genetic information is in everyone's best interests.[20] If such a strategy fails, it might be necessary in the future to evolve new professional protocols in which confidentiality may be guaranteed to families rather than individuals because of the 'familial' character of genetic diseases.[16]

With new genetic knowledge, the information received in genetic counseling will soon be outdated in some situations. Although the 'duty of recontact' is not part of standard care, some consider that responsibility for staying in contact should be shared between health professionals and patients.[21] For practical reasons, the genetic counseling centers cannot always fulfill such a duty to recontact when new, important information emerges. Instead, all families might be encouraged to contact the genetic counseling unit and ask about possible new information whenever they feel the need of updating their knowledge (for instance, before engaging in a new pregnancy).

Genetic counseling must be based on up-to-date knowledge of the disease, and the genetic counselors must have the required capacities to help families to make decisions and to make the best adjustment to their situation.[22,23] Some authors add that counseling should preferably be available in the individual's own language or, alternatively, interpreters should be used.[24] Also, in case of complicated or detailed data, information should always be given in a written form. Nondirectiveness should be emphasized.[25]

In addition to genetic counseling and information given during a personal contact between the counselor and the client, other ways of distributing information to patients and families can be used. These include books, leaflets, videos and websites, and telemedicine approaches. Patients and families can also be informed of existing patient support groups relevant to their problem.

Counseling may appear expensive, as it is time consuming. In addition, genetic tests for rare disorders may, at present, be very expensive. However, as an individual or a family does not need genetic services often, the total cost may not become very high. Genetic counseling can save money as it may help to find the correct diagnosis and stop other diagnostic investigations, it may give information for the family which helps them to cope with the situation, as it may help the physician to treat the disease more adequately.

Prenatal diagnosis As ultrasonographic assessment of embryos and fetuses is becoming a common practice, clinical genetics is now closely associated with the interpretation of antenatal findings, which may reflect a genetic disease, or a severe defect of another origin. Being available for this is part of the duty of care of genetic centers.

Prenatal diagnostic services are offered to families in which there is an increased risk of a disease that can be detected before birth. Again, it is recommended that prenatal diagnostic tests in families with an increased risk of a disease should always be preceded by appropriate genetic counseling and preferably before engaging in a new pregnancy, as well as with sufficient attention for other options like remaining childless, adoption, artificial insemination using donated gametes[26] or the acceptance of having a child with a genetic disorder.[27] Free choice is also emphasized.[28] In addition, policy statements stipulate that the risks and uncertainties of prenatal diagnosis should be discussed with the parents. The prenatal tests must be performed at the appropriate gestational age so that, according to the local legislation on terminating pregnancies, there is enough time for the parents for reaching a decision about the ongoing pregnancy. If the disease is diagnosed prenatally, the family still has free choice either to continue the pregnancy or to terminate it. If the family chooses not to use prenatal diagnostic possibilities, the follow-up of the pregnancy and care for the newborn must happen according to their choices.

In many countries there is a systematic risk assessment for aneuploidy based on maternal age. It has been argued that the concept of risk assessment solely based on maternal age should be complemented by a more integral risk screening concept based on nuchal translucency, ultrasound in general, and maternal serum markers which are already used in many countries.[29] In some countries, all mothers-to-be regardless of their age, are informed about risk assessment for aneuploidy.

Cascade carrier testing Cascade carrier testing, that is, testing along family lines starting from an index case, involves the diagnosis of an affected individual followed by the systematic identification and testing of relatives. However, information of relatives in cascade testing is difficult. Even if the disease is known in the family, information on the genetic risk may only be disseminated to a minority of those relatives at risk, or relatives may refuse to be tested. Using individuals within families to initiate the contacts with distant relatives, which is necessary when cascading goes further than about the first cousin level, is an activity that is likely to make genetic counselors unpopular in the community.[30] There are many problems associated with letting patients inform relatives,

for example, the quality of the information, and the lack of certainty that all relatives are informed.

As for the other types of testing, it is essential that each tested family member gets information and counseling before the test.[31] Also, the result of the test and the explanation of its implications should be given in a written form. Some argue that adolescents might be able to benefit from carrier testing, at their own request, for making reproductive decisions, but carrier testing of children before adolescence should be avoided.[32]

Presymptomatic and predictive testing It has been demonstrated that it is in the category of presymptomatic and predictive testing that most of the difficult issues involving genetic testing lie.[33] It should be noted that the term 'presymptomatic testing' is best reserved for those situations where an abnormal test result will almost inevitably lead to development of the disease at some point in later life, whereas the term 'predictive testing' covers a broader range of situations in which the risk of a disorder occurring is substantially increased or reduced, but without necessarily implying any degree of certainty.[33]

Presymptomatic and predictive testing provide information about the 'future health status' of a healthy person; an asymptomatic person with the mutant gene will stay healthy for an unpredictable number of years. The availability of presymptomatic and predictive testing gives informed people the choice 'to know' or 'not to know', a decision with tremendous consequences.[18]

Presymptomatic or predictive testing is available for two major categories of diseases: neurogenetic diseases and hereditary cancers. Even if the approach for neurogenetic diseases has been a valuable starting point for predictive testing for hereditary cancers, one should keep in mind important differences between testing for 'incurable' neurogenetic diseases and testing for hereditary cancers, for which 'prevention and treatment' may be available. Some consider that the approach should be tailored to each specific disease.[18] For instance, although the identification of the BRCA genes promises a possible future determination and treatment of women and men who are genetically susceptible to cancer, current data reveal certain dilemmas and uncertainties regarding our ability to interpret the results from testing and offer effective management options.

The psychological complexity of presymptomatic and predictive testing requires careful consideration. Information about genetic risks and results of genetic tests has far reaching implications, not only for the counseled or tested persons' own well being but often for their relatives' future too. Therefore, an adequate and systematic multidisciplinary approach as well as ongoing education of professionals and of the general public has been recommended to avoid pitfalls. Proceeding with care and flexibility is also encouraged.[34]

Education and training of primary care providers and non-MDs involved in genetic counseling Although many universities in Europe have gradually improved the official teaching of genetics during the medical schools' courses and during the postgraduate schools' courses, most of the medical geneticists spend a significant proportion of their time on educational initiatives for colleagues or other health professionals. In several countries clinical genetics is officially recognized as a specialty, and many countries have centers where physicians can be trained in clinical genetics. In addition, more and more specialists in different fields of medicine start a training period in a genetics department (or genetic laboratory). This process has been, so far, spontaneous with no official planning and recognizing this activity first at the European level and consequently at the national level appears urgent.

Primary care providers. Scientific advances in genetics point to the need for primary care providers (obstetricians, gynecologists, pediatricians, internists, general practitioners, etc) to develop the necessary skills to assess genetic risk, discuss the implications of genetic testing, and appropriately refer individuals to specialists.[13,35,36] Providers of primary care who are poorly informed can have a detrimental effect on the welfare of patients and their families.[37] The danger that genetic centers will be submerged beneath a flood of inappropriate referrals also exists.[38] The quality of referrals as well as the continuity of care is important. Continuity requires effective means of identifying and following up family members who are at risk. Genetic disorders frequently involve more than one-body system and require multiple investigations in a variety of different specialties, so coordination of clinic visits is essential to avoid unnecessary duplication and major disruption of the patient's life.[38]

In 1990, a study carried out by the Clinical Genetics Committee of the Royal College of Physicians of London found that primary care providers have had little undergraduate genetic education and are unlikely to have had any systematic postgraduate genetic training.[39] Still, 10 years later, a multidisciplinary inquiry into counseling for genetic disorders by nongeneticists showed that poor recording of genetic counseling in hospital records was a common finding and a major impediment to the assessment of the routine genetic practice of non-geneticist clinicians.[40] Therefore, standards for education and training were proposed, such as:

- 'The need for a national policy for auditing and improving undergraduate and postgraduate medical education in genetics should be considered.
- Health professionals in all specialties and in primary care should have training in the basics of genetic management and prevention of disease. All health professionals should know how to obtain a basic family history and when and how to obtain specialist genetic advice.

- Health-care professionals involved in the provision of antenatal care should receive special training in genetic issues.
- The role of primary care must be recognized by enhanced training of community staff in identifying, appropriately referring and providing continuing care for those at risk'.[40]

Initiatives in undergraduate medical education are under way. Primary care providers' knowledge and expertise can also be increased through contacts with specialist centers, special interest groups, use of guidelines, and by collaboration in research projects as well as through the traditional forms of continuing education.[2]

Other healthcare professionals involved in genetic counseling. In Europe, although genetic counseling has been traditionally considered part of the clinical assessment and therefore only clinical geneticists could perform it, during the last 5 years a comparison with the North American organization model of the whole process has started. In North America, genetic nurses and genetic counselors with master's degrees have functioned as members of a comprehensive genetic service for many years; they are trained to collect and confirm medical and family history information, perform risk assessments, offer patient education regarding genetics, and provide supportive counseling services for patients and families.[41,42] In Europe, several centers in different countries (eg, Belgium and The Netherlands) have worked for many years with multidisciplinary teams, consisting of physicians as well as healthcare professionals from psychosocial disciplines.

In September 1996, an International Meeting on 'Psycho-social aspects of genetic counseling,' held in Rome, pointed out the complexity of the consequences of a diagnosis of genetic disease, in particular from the patients' point of view. The need for other professionals such as psychologists, nurses or social workers was underlined in the meeting and also afterwards.[43] In ESHG meetings in 1997 and 2000, satellite workshops on 'Education, Training and Responsibilities of Non-MD Genetic Counselors' and 'Training of non-medical genetic counselors in Europe' were organized. It emerged that an accreditation mechanism is needed for genetic counselors across European countries as well as a 'career' progression.

In the United Kingdom, recommendations for education and training of genetic nurses and counselors have been proposed.[44] They emphasize the core competencies considered essential for practice and acknowledge that these may be achieved by different educational pathways. The guidelines consist of (1) specific knowledge or skill requirements; (2) the educational or training path recommended to achieve them; (3) an appropriate means of assessment.[44] While higher education may equip nurses with a scientific knowledge base and critical thinking skills, empirical learning will assist the development of the professional competency.[45] The psychosocial issues can only be adequately addressed if training has included supervised clinical experience over a sufficient period of time, and where feedback and reflection is sought and given.[44]

The need for well-trained non-MDs appears particularly obvious nowadays. The workload of clinical geneticists has increased more rapidly than their number.

Clinical genetics and other specialties Advising and treating families with common monogenic disorders have traditionally been performed by internists and general practitioners. Similarly, testing for Rhesus incompatibility has been part of maternity care and PKU-screening part of neonatal care. These tasks would have been too laborious for the small number of genetic specialists, and specialists in other fields of medicine have the essential genetic knowledge for these tasks. In the future, there will be a growing number of situations falling in-between clinical genetics and genetics in medicine performed by other specialists. These include predictive testing in common multifactorial diseases if some tests are proved to be useful for managing the disease, carrier screening for recessive disorders and pharmacogenetic testing prior to the administration of drugs tailored to individuals' genetic profiles.

Clinical geneticists have here an important task in planning and supervising such work, in preparing information for the public and in educating the personnel for such projects, including the public health officials. They also have a responsibility to share their knowledge with the other specialists involved in genetic testing. For these reasons, it is important to establish strong relationships between clinical geneticists and specialists in other fields of medicine.[46]

In other respects, many tests have a low predictive value and must be interpreted with caution. Only teams working in a multidisciplinary setting, including molecular geneticists, cytogeneticists, biochemists, and clinical geneticists are likely to provide an accurate interpretation of the test results.[47,48] It appears essential to an understanding of genetic testing to distinguish in common complex diseases, rare subsets of disease due to single genes of high penetrance from susceptibility genes of low penetrance.[16] Testing for low penetrance susceptibility genes has still a weak clinical utility: clinicians are not able to predict outcomes for their patients as well as public health physicians for populations.

Organization

Advances in the understanding of genetics and of molecular biology and the translation of that knowledge into health care will have a profound effect on the future organization and delivery of services. Increased interest and demand from patients and practitioners is already being felt, especially where genetics centers are developing

new services and raising awareness and expectations, as well as for commercial reasons. Awareness is even greater in the United States, where websites and private genetic centers promote genetic issues and testing opportunities.[36]

Education and training of clinical geneticists In most countries the specialty of clinical genetics has been officially recognized. The requirements for specialization are in the average: service in clinical genetic units for 3–4 years, in a molecular or chromosomal laboratory for 1–2 years, with either of these services including a period in prenatal diagnostic centers. In countries where clinical genetics is not a specialty, it has been proposed that a physician should work as senior physician in a clinical genetic unit only if he/she has experience in clinical genetics of the same order as that required for the specialty in the other countries.[1] In 2001, the European Society of Human Genetics recommended a *Formal recognition of medical genetics as a medical specialty in Europe*.

In addition to initiatives to develop medical schools' curricula so that genetics education is fully integrated, there are many web-based and CD-ROM resources for continuing education programs. These resources aim to promote the appropriate use of genetic counseling and genetic testing in patient care. They provide reliable, easy-to-use and current genetic testing information for the benefit of families and their health-care providers. Given the fast pace of scientific research in medical genetics, much of the up-to-date information that will be required to practice clinical genetics most likely will reside on the Internet.[49] Although no single Internet site can provide comprehensive information, an amazing depth of content already is online, such as databases on human genes (human genome resources, OMIM, GDB, Genatlas, Genecard), on mutations (HUGO), on phenotypes and protocols (OMIM, Geneclinics, Pedbase, Orphanet, Nord), as well as databases on services (labs, specialized clinics, patients' groups, research projects, clinical trials). However, online genetics resources have other potential effects: (1) an increase in referrals for both clinical and research testing; (2) a possible over consumption of tests or services; and (3) advertisement for unreliable services. Thus, codes of conduct for online genetics resources imply transparency, an absence of conflict of interest, an updating process, a quality chart, an editorial committee with a peer-review system as well as interactivity. Codes of ethics for online genetics resources must be strict in order to protect the users.

Providers of services All clinical specialties use genetic techniques and approaches in their practice; but the report prepared by the Royal College of Physicians of London[11] showed that specialists in other fields and primary care physicians may feel unable to handle genetic issues and developments and may expect clinical geneticists to do this for them, giving a need for greatly increased numbers. Signs of this are appearing in referrals for familial cancers.[42] Cardiology provides an example of a specialty where genetics involvement may become as extensive as is currently the case for cancers. According to the Council of Regional Networks for Genetic Services (USA),[11] clinical guidelines are needed to define clearly the quality of care delivered by genetic services providers. In addition, primary care providers as well as specialists may need guidance if they have to participate in the initial work-up and ongoing management of patients and families affected by or at risk for genetic diseases.[35]

In each European country, CAGSE has recommended that there should be clinical genetic units providing specialist genetic services for the families.[1] The units should not be too small and should have a possibility to exchange their experience and consult with one another. Because of the wide range of – rare – genetic disorders, collaboration between professionals is essential. In fact, rare diseases are not part of every physician's experience or training, many syndromes only manifest fully over years, or continuous follow-up of chronic patients often does not happen. Collaboration with families and patients' organizations is also crucial because they often have a long-time experience, medically and socially, about the disease, which otherwise is missing among so-called experts. Thus, there is a strong need for developing international networking for rare diseases in order to have professional networks, easily readable overviews of rare conditions, bioinformatics resources, but also interactive workshops presenting unresolved cases, as well as publications presenting unresolved, puzzling cases.

Many consider that the clinical genetic services should have multidisciplinary teams composed of MDs and non-MDs, including several psychosocial disciplines.[2,18] Usually, the core team of a genetic service is composed of clinical geneticists and nonmedical genetic counselors, with access to psychologists, social workers and other medical/nurse specialists. Nonmedical genetic counselors have been trained in human and clinical genetics and counseling, sometimes in formal programs but sometime only in supervised practice. Clinical responsibilities are shared between clinical geneticists and nonmedical genetic counselors, with the exception of diagnosis, discussion about prognosis and management of the disease and further diagnostic investigations that are the responsibility of clinical geneticists. Nonmedical genetic counselors have an important role in supportive counseling and reinforcement. Teaching responsibilities are also shared between clinical geneticists and nonmedical genetic counselors. The latter have a special responsibility for designated general practices and specialty clinics, and constitute a point of access to centers.

Finally, clinical units must closely collaborate with diagnostic laboratory facilities. The organization of genetic

laboratory services has been greatly improved over the last decade, whether for increasing diagnostic resolution or mutation detection techniques or bioinformatics.[13] Thus, it is strongly suggested that the organization of clinical genetic services should optimize the provision of cytogenetic, metabolic, and molecular genetic testing services, as well as the collaboration with university hospitals and with human genetic research centers.[12,50]

The requirement of well functioning units of sufficient size is so important that it can override the patient's possible wish to have a short way to the clinic. However, the patients' desires to have a short way to the clinic should not be underestimated. Geographical inequalities exist everywhere, with rural people getting fewer services. Outreach services like visiting nurses, telemedicine, or internet may be envisaged. Also, the socioeconomic situation and level of health care services differ among the European countries. The assortment of specialist genetic services is very wide and it has been anticipated that all European countries will not be able, in the coming years, to offer all these services to their inhabitants.[51]

Regional centers* vs *local centers There are no reliable reports on the organization of genetic services in each country, except for the CAGSE report (1997), which showed that the organization models are different. Models for service development have primarily been set up by genetics centers alone or in collaboration with academic departments of general or family practice, or with health maintenance organizations (HMOs) in the United States, or with individual family physicians. In the United States, genetic centers now offer comprehensive genetic care plans for large HMOs or to other providers and their patients.[52] In Europe, not all centers operate in the same manner, reflecting the differing geographical locations served or the different facilities depending upon their resources and those of their nongenetic colleagues.

For some rare diseases, national and supra-regional services have been established. Since a large number of genetic diseases are rare with a very low frequency among population groups, it is unrealistic to imagine that local laboratories could meet the future genetic testing demand for the enormous number of rare genetic diseases. The availability of genetic testing for these diseases will more likely be dependent on using cross-European genetic testing services provided by specialized reference laboratories. However, this should always be associated with appropriate genetic counseling in the patients' own country.[1] Other centers undertake disease-specific clinics and actively manage the surveillance for complications in specific diseases. Such clinics would not be a 'core service' but may form part of the agreed protocol of care for that center.[11,53]

Genetic services in many countries have been developed as multidisciplinary regional centers with strong links with academic human genetics. Close collaborations between regional centers and professional groups has resulted in the establishment of a number of highly effective national networks – for instance, in oncogenetics and neurogenetics – to ensure that particular expertise is utilized for the benefit of patients from any region. Such networks are considered very useful. Finally, recognizing that clinical and laboratory services need close links, coordination in purchasing of all elements of regional services has also been promoted.

Genetics in multidisciplinary teams Considerable expertise has been built up in the delivery of clinical and laboratory services for a wide range of genetic disorders. Such expertise can be used for the development of services for further conditions, as research and technological advances allow. Effective working relationships have been established with various clinical specialists as well as with those in primary care. However, joint appointments have been rare. With the growth of genetic applications in specific fields, joint appointments offer opportunities that may allow the necessary clinical specialist and genetics expertise to be combined. Joint appointments between oncology and genetics centers are an example that has evolved in several regions, and this pattern might be seen in other specialties.[11]

The important points to highlight in developing genetics in multidisciplinary teams are: (1) the need for recognition of professionals involved in genetic counseling, and ensuring the proper training and supervision for such professionals, as well as developing a recognized career pathway for them; (2) the need to find their role not only in counseling rare hereditary diseases but especially in more common diseases, for instance, families with suspicion of hereditary cancer and other immoderate risk individuals;[54,55] The development of such services requires that responsibilities of different disciplines need to be understood and agreed upon.[45] Such a system has been developed for hereditary cancers in Finland, where a three-level model of collaboration has been instituted, involving nurses, primary care physicians and medical geneticists. Specially trained nurses[56] working in the regional cancer societies have preclinic responsibilities; when the situation requires consultation with a physician, primary care physicians document in more detail the cancer history and then, when needed, medical geneticists do genetic counseling, testing and follow-up. Although this three-level model still needs to be improved and assessed, it could be suitable for other common disorders, for instance dementia. In addition, genetic counselors and genetic nurses have important and often rather independent roles in prenatal screening programs and other situations where counseling is needed for a large number of clients.

As the specialties continue to expand, members of departments will need to work in a collective manner so

that most of the functions relating to patient care can be delivered to the population that they serve. For instance, in prenatal genetics, (safe) invasive procedures, (reliable and rapid) laboratory testing and (reliable and rapid) risk assessment require the collaboration of obstetricians, geneticists, and clinical chemists. Prenatal genetics requires early decision making, alternative options, as well as rapid definitive results. Ideally, to help parents in their decision-making, organizations should strive to shorten delays so as to afford parents as much time as possible to weigh their options when faced with unexpected lab results.

Collaborating by dividing up the work is a practical approach allowing clinical geneticists and other specialists to share their experience, to start specific programs, protocols for specific conditions, or problem/patient-based approaches. But such collaboration takes mutual respect and honesty to collaborate as professionals; it is also only possible in multidisciplinary situations and it involves financial and other practical aspects.

Moving from research to clinics Service development is a crucial part of genetic service provision and maintenance of quality, given the rapid changes. Mechanisms need to be developed to translate beneficial research findings into service in a framework, which allows for evaluation and further development. Patient support groups who collaborate in research want to be reassured that once the research is over the service will continue if proven to have benefits.[57]

Undoubtedly, the ability to document the genetic structure of individuals will allow a greater understanding of disease mechanisms and a better prediction of their susceptibility to disease, but the pace at which these risk predictions will become useful in the clinical situation, or the extent to which individuals will change their behavior or allow effective interventions to alter that risk, is uncertain.[16,47] Also, the new technologies associated with genetics and the increasing expectations of patients may create significant cost pressures that will have to be carefully considered in the context of health-care systems with finite resources at their disposal.[16,58]

Several factors must be considered in the decision whether new genetic tests and new medicines should be part of service provision. Sufficient attention must be paid to the elaboration of the appropriate counseling context in a genetic center, preferably involving a multidisciplinary team. For some other tests the training of other physicians may be necessary. In the latter case, an area of concern is whether physicians are able to interpret results for their patients.

While the large potential markets for genetic tests for common conditions make these tests attractive to commercial concerns, marketing such tests raises questions. One such question is whether the patient actually benefits from knowing his or her risk of disease. Another question is that physicians may have little to offer in terms of preventive and curative strategies. It is recommended that adequate standards should be set for all aspects of genetic testing, both in relation to analytical validity and clinical utility. It might take many years to be sure that the identification of groups at risk for common diseases, or for specific drug therapies and consequent interventions, is beneficial. It is generally considered that major programs of for instance genetic screening should not be launched until the scientific basis of specific genetic developments is beyond doubt.[2,16,59,60] The danger is of inadequately evaluated, unregulated tests of potential promise but unproven value being passed on to service providers.

Public* vs *private In Europe, genetic services are mainly paid for by the public health care system and equally available for all citizens independent of their economic or insurance status.[2] If an individual wants extra services, for example, carrier testing for rare recessive disorders for which they have no increased risk, such services are provided in some countries by private health-care system at their own expense.

For some laboratories the number of tests performed may be of great interest. Pressure to maximize testing may influence the way information is presented, and there is a potential danger of testing being encouraged, regardless of individual benefit. Commercial testing has already been launched for a number of different genetic disorders. Genetic testing services can be offered to the public with minimal genetic counseling. In the United States, there is a growth of private genetic services that are marketed directly to the public.[36,61] Another difficulty appears if a private company is able to obtain a patent on a particular gene. The most publicized example is the patenting of the BRCA1 gene. This has enabled a company to specify terms on which other laboratories may, or may not, test for mutations in the gene. In the USA, other laboratories are not permitted to test for BRCA1 and BRCA2 mutations, and some might have lost important expertise and manpower as a direct result.

This raises questions about whether health-care systems should be obliged to pay increased amounts for each test done by the company, thus reducing the number of tests that they may be able to offer within the health service budget, and introducing the possibility that a two-tier health-care system could develop, where some private individuals may be able to pay the fee for the test if the health service cannot afford it in their case. In addition, there is the question of who provides and funds the genetic counseling for the privately performed tests.[62] An unscrupulous approach could ruin public confidence in genetic testing and give responsible companies a bad name. Regulations protecting the public and setting standards for industry could therefore be of great benefit to all parties

involved.[58,63] The challenge is how to maintain standards as testing encompasses increasing numbers of diseases and pressures to contain costs of genetic services increase, particularly since the cost of pre- and post-test activities may equal or even exceed the laboratory costs of the test itself.[64]

Funding In recent years, the workload of clinical genetic services has increased dramatically. But the increasing workload has not been paralleled by increasing resources, and there is considerable variation among regional centers in terms of staffing and other resources. The pressure on clinical genetic services is increasing as knowledge advances, as the possibilities increase (in particular, if there are significant breakthroughs in pharmacogenetics) while new resources fail to keep up with progress.[65,66] It has been argued that priorities will need to be set even with projected increases in the level of funding.[16]

However, there is a consensus in order that the costs of specialist genetic services be collectively covered by the public health-care system, health insurance or other means used in the country concerned. The costs should not be left to the individual family and it may be unfair to leave them to a local small community. In the UK, a model of contract for a clinical genetic service with the NHS shows that it is economically feasible.[67] The Belgian system, whereby a decree regulates the organization of genetic services (see Appendix A) can also be considered as an alternative. In the Netherlands, genetic services are incorporated in the health care and funded in such a way that equal access is guaranteed (see Appendix A).

Quality assessment

Quality assessment of clinical genetic services Measuring the quality of services like diagnosing rare disorders or counseling is extremely difficult. In addition it is difficult to outline the requirements of 'minimal' or 'optimal' quality in clinical genetics.

Means of maintaining high standards of quality of clinical care comprise a system of clinical audit in place to assess the use of protocols of care, the accuracy of diagnosis, of pedigree analysis, of risk assessment, the quality of record keeping as well as the quality and promptness of explanatory letters to referring clinician and patient. Means of maintaining high standards of quality of clinical care also comprise a system for active follow-up of investigations: agreed plans for follow-up of patients and notification of kindred, support, including the arranging of prenatal tests and post-termination counseling.[11] It has been suggested that each clinical genetic unit should collect standard data on which its effectiveness can be measured. They should include: (1) the numbers of families/individuals seen; (2) districts of residence of patients and family members; (3) number and type of individual diagnoses; (4) number of patients seen at central/peripheral clinics; (5) sources of referral: general practitioners, pediatricians, obstetricians, other consultants, nurses, self-referrals, other; and (6) measures of the amount of benefit for the patients, such as the 'Perceived Personal Control'.[11,40,68,69] The Health Council of the Netherlands[20] also proposed regulations on clinical genetic testing and counseling in order to promote high quality standards. The committee concluded that concentration of clinical genetic testing in university centers has contributed to continuity and quality improvement; this concentration should be maintained in view of the nature of genetic counseling. In the light of recent developments in the field of clinical genetics, the committee recommended that the professional groups involved in clinical genetics should have responsibility for drafting and updating quality requirements; in this context, the government's role should be supervisory.

Quality of counseling process may also be measured. In genetic counseling, probability information is essential; the context and the presentation of risk information influence the subjective perception of the information and the subsequent decision.[70] Clinical geneticists 'are said to espouse a nondirective' method of counseling clients, but frequent deviations from non-directiveness have been shown.[19,71] Another approach called 'shared decision making' has recently been introduced into genetics and seems to fit well into some counseling situations.[72] The meaning of 'nondirectiveness' has been extensively discussed and the definition that has been proposed for the genetic context is the following: 'nondirectiveness describes procedures aimed at promoting the autonomy and self directness of the clients'.[73] In this context, a clear view on all options and all outcomes is very important for the clients.[68,70]

In other respects, some aspects of genetic counseling are not reflected in all definitions of genetic counseling and do not seem to have always received sufficient attention in practice and training. There are two basic approaches to genetic counseling: (1) the teaching model and (2) the counseling model. Attempts are made to combine a teaching model with a counseling procedure. However, the skills needed for teaching and counseling differ so vastly that it has been claimed to require 'unusually gifted and flexible' professionals combining them both. It is essential that both pedagogical and counseling skills, including the awareness of one's own biases, should receive sufficient attention during training programs. On the other hand, a team approach by professionals from different disciplines – medical, psychological, social – also contributes to the combination of a teaching model with a counseling procedure.

Evaluating the success and quality of genetic counseling is complex not only because of the different approaches to genetic counseling, but also because it is difficult to define adequate outcome measures that are compatible with the

aims of genetic counseling. How well do patients recall information? How has genetic counseling altered their plans? How have they in practice chosen to act on the basis of the information received? How satisfied are they with the process of genetic counseling? Information, reproductive plans and reproductive behavior cannot be considered as simple numerical measures of success or effectiveness of genetic counseling. Thus, some argue that a typical audit-approach for assessing the success or effectiveness of genetic counseling is not feasible and could give rise to misleading conclusions and wrong decisions in the allocation of resources for health-care and clinical services. Perceived personal control seems to be an appropriate outcome measure. This concept was found central to coping with health threats and to adapting to a broad spectrum of health problems. In a study of genetic counseling cases, comparisons of the perceived personal control scores, before and after counseling, showed significant increases of perceived personal control.[68] A 'retrospective assessment of satisfaction' approach is another appropriate outcome measure. Indeed, relying on statements of satisfaction of 'naïve' clients may be inadequate. They are in a much better position for this type of judgement some time after the process of genetic counseling is completed. Simple outcome measures seem attractive but give a too superficial, incomplete picture of the quality of genetic counseling. Yet, global outcome measures for the entire process of genetic counseling seem unattainable and, moreover, 'attempts to devise such a measure could lead to inappropriate efforts to reshape the activity of genetic counseling'.[33]

Involving the patients in the quality assessment of genetic services may help clinical geneticists offer a higher quality service. Geneticists want to provide the highest quality service. But what are the patients' expectations? Patients' concerns may differ from geneticists' concerns, and any outline of optimal services should reflect both. This objective has implications for the development of ways to build public confidence in, and understanding of, medical genetics. In the absence of this dialogue, there is likely to be both unrealistic optimism and unrealistic fear about genetic services. Links with the patients or their representatives is also fundamental in achieving full and equitable delivery of genetic services, since major problems in service delivery are perceived by patients and families. Access is not always consistent or satisfactory, services are not always adequately linked to other medical specialties, nor are all aspects of patient situation(s) necessarily addressed.

Finally, when considering the quality assessment of clinical genetic services, it is necessary to consider how the broader teaching of genetics is organized and how it becomes an integral part of all the medical specialties. Teaching and training in clinical genetics and genetic counseling are still strongly needed. In the last 10 years, several countries have recognized genetics as a medical specialty. However, very few European countries have specific regulations regarding teaching and training in medical genetics. Only in the United Kingdom there is a long history of regulation of medical genetics training. At the European level, various suggestions have been made, such as (1) to develop a common curriculum and training activities for postgraduate programs in medical genetics; (2) to recommend the creation of national bodies board of clinical genetics, specialty advisory committees, specialist registers, and accreditation systems; and (3) to improve the international exchange of experiences (European courses, practical short-term training periods in a different country, an accreditation system for participating in European quality-control studies).

Quality assessment of genetic laboratory services The laboratory procedures in genetic laboratories and/or centers should be under similar quality control as the other clinical services. High-quality laboratory services include the identification of laboratories and units with which the clinical genetic unit can connect and the participation of those laboratories in a quality assurance scheme. An attempt to coordinate External Quality Assessment systems has been developed through the European Molecular Genetics Quality Network, the European Concerted Action in Cystic Fibrosis and the UK National External Quality Assessment scheme for molecular genetics, which includes Ireland and the Netherlands.[74–76] External quality assessments for testing strategies have been organized in different diagnostic laboratories for different diseases (HD, CF, Y chromosome deletion) and laboratory guidelines have been proposed. A proper implementation/validation of testing as well as quality controls and quality assessments have been recommended. The present molecular diagnostic situation is in fact far from ideal: laboratory errors are made; in other respects, it is difficult to get additional funds for the implementation of diagnostics or new technologies, in spite of a greater demand from clinicians and patients, there are many genes to be tested and many new emergent technologies.[74] Training programs for staff members and laboratory accreditation by professional bodies are also needed.

Many genetic diagnostic laboratory services are organized in close – physical or functional – contact with a clinical genetic service. In the not so distant future, common technical platforms may be created for the diagnosis of genetic and of acquired (malignancy, infections) diseases as well as for the genetic predispositions and for pharmacogenetic polymorphisms. It has been argued that geneticists should discuss with their colleagues (clinical chemists, pathologists, microbiologists, and medical specialties) on how to organize the laboratory activities in the future. At the least, a genetically trained MD with a long experience in clinical genetic services together with

one properly trained non-MD could be partners of the team that will run these technical platforms in the future. Several geneticists are concerned that genetic testing, becoming more kit-based and more automated, continues to be provided in an adequate counseling framework. They are also concerned in retaining the confidence of the public in genetic testing by promoting and improving standard of quality in all the centers involved.[13]

The American College of Medical Genetics[77] developed standards and guidelines for clinical genetic laboratories. These standards and guidelines aim to assist medical geneticists in providing accurate and reliable diagnostic genetic laboratory testing consistent with currently available technology and procedures in the areas of clinical cytogenetics, biochemical genetics and molecular diagnostics. In determining the propriety of any specific procedure or test, the medical geneticist should apply his or her own professional judgment to the specific circumstances presented by the individual patient or specimen. Regarding personnel policies, a laboratory director and/or technical supervisor must have an appropriate doctoral degree and at least 2 years of postdoctoral training and/or experience in his/her clinical laboratory subspecialty and certification or eligibility in medical genetics. Certification in pathology or clinical chemistry with appropriate training and/or experience in genetics may substitute for certification in medical genetics. In addition a clinical consultant is required for all laboratories and (s)he must be a clinical geneticist or a clinical cytogeneticist.

Public education

Today, access to information is easy, in quantities unimaginable, and keeping up to date is a difficult task. Access to the internet exposes the public to genetic information, which can serve to inform as well as to confuse. Also, because of the wide range of rare genetic disorders, it is unrealistic to expect that all primary care providers will be able to help families with rare conditions. Clinical geneticists are often asked to provide talks, lectures, and seminars. Clinical geneticists recognize that they have responsibilities in the area of education, in particular to translate some of the complex scientific concepts inherent in genetics into more understandable information. But clarification of misinformation can be extremely demanding of time. It is now proving almost impossible to respond to all requests, and therefore clinical geneticists must agree together with specialist colleagues in respective departments, genetic nurses, and patient support groups as to how to prioritize the educational demands and how to respond. In the United Kingdom, it has been reported that many departments are involved in developing courses for different patient support groups. In other European countries (eg, Belgium and The Netherlands), genetic centers have played an important role in the development of educational tools (eg, leaflets, videos); in 1999 in Belgium, the Advisory Committee on Bio-ethics organized a large-scale conference on 'Heredity and Society' with the cooperation of members of several genetic centers on the organizing committee as well as during the meeting.

It has been recommended that governments should set an open and wide-ranging agenda for discussion about current and future developments in genetics.[16] There should be a concerted campaign to raise public understanding and awareness. If this process were to be properly managed there is no reason to suppose that genetics should not continue to command wide support among the public.

Telecommunications and the internet will increasingly bring changes to the relationship between physicians and patients, and between patients and the new kinds of knowledge to which they have access. These technological developments and trends will affect the future impact of genetic science on health care. For instance, recording and accessing genetic data for a patient will be of increasing importance, in the context of increasing opportunities for prenatal diagnosis, and for prediction of common disease risk. Coupled with referral guidelines and computer decision support, the new communication technologies may provide useful methods of supporting primary care providers in the field of medical genetics; but this will require the information to be in a format that is appropriate for the individual user.[48,78]

Partnership with patients support groups

Some consider that the involvement of service users should be encouraged; service users' concerns may differ from professionals' concerns and any outline of optimal services should reflect both.[79,80] Where umbrella organizations exist, their views will be most helpful in commissioning genetic services. For instance, in the United Kingdom, the Genetic Interest Group (GIG), an alliance of charities and voluntary groups for families affected by genetic disorders, has been running a systematic awareness-raising program, designed to fit in with local patterns of service provision and incorporating continuing medical education recognition.[66] GIG also produced a cross-curricular teaching pack, for use by science and humanities teachers working with adolescents, as well as an information booklet for the brothers and sisters of children in whom a genetic disorder has been diagnosed. This booklet is designed to help parents and professionals for addressing the questions, which raise themselves in the minds of children when a genetic diagnosis is made.[66] In Belgium and The Netherlands, the cooperation between genetic centers and umbrella patient organizations has also resulted in good information tools. In countries without umbrella organizations, the partnership with patient support groups is less organized and usually works well in case of some patient support organizations but fails in case of some others.

Multicultural aspects

Within any country there exists a diversity of cultures and opinions about a number of issues relevant to genetics. These include human reproduction issues and community and individual approaches to the significance of disabilities.[81–83] This diversity has been almost always respected and this should continue to be before setting program goals. However, medical services for ethnic minority groups are targets for improvement. In the United Kingdom, the multidisciplinary inquiry into counseling for genetic disorders by non geneticists found marked regional inequalities of access to genetic services for beta thalassemia major, a disease predominantly of ethnic minority groups.[40,84] These inequalities contrast with what has been shown to be possible in well-documented and successful models of routine thalassemia prevention in Mediterranean populations.

Community meetings may be encouraged to discuss important issues, such as the value of learning about one's own medical family history and where to go for genetic counseling. It has been recommended that future development of genetic services should emphasize the provision of linguistically and culturally appropriate resources.[51] Providers of services require training, resources and information about the relevant groups within their referral area. Some consider important that culturally sensitive issues, such as genetic counseling, for instance, be dealt with by the personnel who speak the patient's language, understand and empathize with the community and relate to its culture. The caregiver should be seen to relate to the culture by the individuals being counseled. Links may need to be established with specialist services in case of diseases such as hemoglobinopathies. Minority ethnic individuals may be involved in both the development and delivery of genetic services for their community.[85,86]

Conclusion

Some general conclusions were identified from the ESHG workshop to arrive at outlines for optimal genetic services. Participants were concerned about equal accessibility and effectiveness of clinical genetic services, quality assessment of services, professional education, multidisciplinarity and division of tasks, as well as networking.

Genetic services are under considerable pressure in order to integrate the new discoveries and to ensure equal accessibility. Patients' needs have increased, whether it be for differential diagnosis for individuals with a manifest disease, predictive testing for at-risk individuals, carrier testing, prenatal diagnosis for relatives of diseased individuals, or different other services for subpopulations. Methods to assess genetic constitution, whether at phenotypic or genotypic level, are routinely employed by many specialties. Genetic counseling is much more than informing a patient or family about the genetic nature of a condition. Thus, it requires specialist training, whether with a MD background or not, and must include a 'psychosocial awareness component'. Setting-up a genetic service requires to provide a basic understanding of medical genetics in undergraduate teaching and educational competence for postgraduate training in medical genetics within other specialties. It requires to appeal to the authorities to grant exclusive educational competence in medical genetics and to mandate formal training in medical genetics to all those participating in a genetic service. Structuring a comprehensive genetic service also requires a team approach, with interdependent specialists and overlapping skills. However, specialties have to be delineated more accurately: the complexity of the team approach is in the coexistence of overlaps and boundaries of responsibilities. Health planners must be aware of the delineation of specialties and stimulated to implement the organization of autonomous genetic services in hospitals that still lack those services.

It is important to prepare all those who work within the health services for the anticipated changes to clinical practice and the consequent demands these will make on services' organization and funding. Of course, the manpower and resource implications for such a wide-ranging educational program could be enormous. Consideration would have to be given to how this huge amount of training and education would be undertaken. The new technologies of information can be used to better inform health-care providers and the public about the possibilities and limits of what genetics can provide to improve the diagnosis and management of genetically determined conditions. Standard protocols will be available soon as well as information leaflets to complement the information provided at individual clinics. Telemedicine will help getting advice from highly specialized centers for very rare disorders. Patients will be able to share their experience with other similarly affected people through internet. This is likely to improve the quality of services delivered and the satisfaction of consumers, this in addition to the further elaboration of the necessary services at the regional level.

Finally, within European countries, adherence to the organizational principles of prioritization, regionalization and integration into related health services would maximize equal accessibility and effectiveness of genetic actions. There is a need for harmonization of the rules involved in financial coverage of DNA tests in order to make these available to all Europeans, regardless of their financial status and origin. Clear guidelines for best practice will ensure that the provision of genetic services develops in a way that is beneficial to its customers, be they health professionals or the public, especially since the coordination of clinical, laboratory and research perspectives within a single organizational structure permits a degree of coherence not often found in other specialties.

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Appendix A

International and national regulatory frameworks

International Organizations *World Health Organization, Report on Community approaches to the control of hereditary diseases, Geneva, WHO, 1985*

This report is concerned with the community aspects of genetics services. It seeks to illustrate their relevance for health care by addressing some quantifiable examples of the control of hereditary diseases; important new technical developments; approaches that may be incorporated into primary health care; evaluation of community-based services; gaps in the existing medical structure that need to be corrected in order to deliver these services; the importance of genetic information in health education; the ethical problems associated with genetics services; and research needs and opportunities.

World Medical Association Statement on Genetic Counseling and Genetic Engineering, 1987 (http://www.wma.net/e/policy/17-s-_e.html)

The World Medical Association adopted this statement to assist physicians with the ethical and professional issues that raised from scientific advances in the field of genetics. In regard to genetic counseling, the Association stated that 'physicians who consider contraception, sterilization and abortion to be in conflict with their moral values and conscience may choose not to provide genetic services. However, in appropriate circumstances, the physician is nevertheless obligated to alert prospective parents that a potential genetic problem does exist, and that the patient should seek medical genetic counseling from a qualified specialist'. Whether physicians advocate or oppose providing the above-mentioned services, they 'should avoid the imposition of their personal moral values and the substitu-

tion of their own moral judgment for that of the prospective parents'.

World Health Organization, Community Genetic services in Europe, Geneva, WHO, 1991

This report gives countries the necessary information to start the rational planning of genetic services based on the assessment of needs.

World Medical Association Declaration of the Human Genome Project, 1992 (http://www.wma.net/e/policy/17-s-1_e.html)

The World Medical Association recommends that 'The genetic service should be easily accessible to everyone in order to prevent its exploitation by only those who have resources which will increase social inequality. There is a need for international information and transfer of technology and knowledge between countries'.

World Health Organization, A Declaration on the Promotion of Patients' Rights in Europe, Geneva, WHO, 1994 (http://www.fgov.be/WHI3/per…onths/wwhv2n1tekst/WWH19019804.htm)

This document sets a series of principles for the promotion and implementation of patients' rights in WHO's European Member States. Under the first principle, 'Human rights and values in health care', it is stated that 'everyone has the right to the protection of health as is afforded by appropriate measures for disease prevention and health care, and to the opportunity to pursue his or her own highest attainable level of health' (Principle 1.6). The second principle on 'Information' stipulates that 'information about health services and how best to use them is to be made available to the public in order to benefit all those concerned' (Principle 2.1). The fifth principle regarding 'Care and treatment' establishes that 'everyone has the right to receive health care as is appropriate to his or her health need, including preventive care and activities aimed at health promotion. Services should be continuously available and accessible to all equitably, without discrimination and according to the financial, human and material resources which can be made available in a given society' (Principle 5.1). According to Principle 5.2, 'patients have a collective right to some form of representation at each level of the health care system in matters pertaining to the planning and evaluation of services, including the range, quality and functioning of the care provided'. Finally, 'patients have the right to a quality of care which is marked both by high technical standards and by a humane relationship between the patient and health care providers' (Principle 5.3).

World Medical Association Declaration of the Rights of the Patient, 1995 (http://www.wma.net/e/policy/17-h_e.html)

The World Medical Association considers that physicians and other persons or bodies involved in the provision of health care have a joint responsibility to recognize and uphold the principal rights of the patient. In the context of biomedical research, the human subject is entitled to the same rights and consideration as any patient in a normal therapeutic situation. Consequently, in regard to the 'Right to medical care of good quality', The Association recommends that 'Every person is entitled without discrimination to appropriate medical care. (…) Quality assurance always should be a part of health care. Physicians, in particular, should accept responsibility for being guardians of the quality of medical services' (Principles 1a–1d). Principle 9 on 'Right to Health Education' states that 'Every person has the right to health education that will assist him/her in making informed choices about personal health and about available health services. The education should include information about healthy lifestyles and about methods of prevention and early detection of illnesses. The personal responsibility of everybody for his/her own health should be stressed. Physicians have an obligation to participate actively in educational efforts'.

World Health Organization, Control of Hereditary Diseases, Technical Report Series No. 865, Geneva, WHO, 1996

This report offers advice on the organization of genetic services in industrialized and developing countries alike, and discusses the ethical, social and legal aspects of genetic technology in medicine, concluding that the broadest ethical issue in the area of genetic services is their limited availability.

World Health Organization Proposed International Guidelines on Ethical Issues in Medical Genetics and the Provision of Genetic Services, Geneva: WHO, 1997 (http://wwwlive.-who.ch/ncd/hgn/hgnethic.htm)

The proposed guidelines are designed to assist decision-makers at both national and international levels to protect people and families with genetic disabilities, to recognize the great potential of advances in human and medical genetics for public health, and to develop policies and practices that will ensure that these applications can become accessible to all and are provided with due regard to ethics and justice worldwide.

The issues related to ethics and the provision of medical genetic services are the following:

(1) General ethical considerations: 'The medical application of genetic knowledge must be carried out with due regard to the general principles of medical ethics'.

(2)The proper use of genetic data: 'It is ethically imperative that genetic data should only be used to the advantage of members of a family or ethnic group, and never to stigmatize or discriminate against them'.

(3) Voluntary use of genetic screening and testing: 'Every genetic test shall be offered in such a way that individuals and families are free to refuse or accept according to their wishes and moral beliefs. All testing should be preceded by adequate information about the purpose and possible outcomes of the test and potential choices that may arise. Children shall only be tested when it is for the purpose of better medical care, as in the case of newborn screening when early treatment will be of benefit to the child'.

(4) Prenatal testing: 'Prenatal diagnosis should be offered to those who need it, but there must be no pressure on couples to accept such testing, nor to use the results of the test to compel either continuing or terminating a pregnancy when the fetus is affected with a genetic disorder. (…) Prenatal diagnosis should be done only to give parents and physicians information about the health of the fetus'.

(5) Justice demands equitable access to services: 'Genetic services for the prevention, diagnosis and treatment of disease should be available to all, without regard to ability to pay, and should be provided first to those whose needs are greatest'.

(6) 'Genetic data should be treated as confidential at all times'.

(7) Genetic counseling: 'Counseling is essential before any genetic testing is carried out, and should continue afterwards if the results entail choices for the person and family tested. Genetic counseling should be available to all, and should be as non-directive as possible'.

(8) 'Education about genetics for the public and health care professionals is of paramount importance. (…) It is important that education about genetic principles relevant to human health be emphasized appropriately for all people in all cultures. Education is a two-way process, and geneticists and other health care professionals have much to learn from support and advocacy groups representing those with genetic disorders. Such groups are an integral part of genetic services, and should be guaranteed a voice in policy and education'.

United Nations Educational, Scientific and Cultural Organization, The Universal Declaration on the Human Genome and Human Rights, 1997 (http://www.unesco.org/ibc/uk/genome/project/index.html)

The UNESCO Declaration is the first international normative instrument in the field of bioethics. Article 5 states that '(a) Research, treatment or diagnosis affecting an individual's genome shall be undertaken only after rigorous and prior assessment of the potential risks and benefits pertaining thereto and in accordance with any other requirement of national law'.

World Health Organization, Medical Genetic Services in Latin America, Report of a WHO Collaborating Center for Community Genetics and Education, 1998 (http://whqlibdoc.who.int/hq/1998/WHO_HGN_CONS_MGS_98.4.pdf)

This document is not a formal publication of the WHO. In the framework of the 9th International Congress of Human Genetics in 1996, a group of experts in medical genetics from Latin America discussed the situation of medical genetics in the Region and set forth a series of recommendations for the continuing development of the field in the areas of services, training and research.

World Health Organization/WAOPBD, Services for the Prevention and Management of genetic Disorders and Birth Defects in Developing Countries, Report of a joint WHO/WAOPBD meeting, The Hague, January 1999 (http://www.who.int/ncd/hgn/reppub_malta.htm)

This document is not a formal publication of the WHO. An Advisory Group constituted mostly by geneticists from 13 developing countries was convened on January 5–7, 1999 by the World Health Organization and the World Alliance of Organizations for the Prevention of Birth Defects, to address the lack of genetic services in the developing world and make recommendations for their growth. Its main recommendations are: need that health professionals and public health officials of developing countries recognize the burden imposed by birth defects and genetic disorders; need for political will and commitment for their prevention and management; define goals of genetic services in terms of individual and family well-being as well as of public health; improve reproductive health, prenatal and newborn care with particular attention to maternal age, nutrition and teratogen avoidance; organize comprehensive genetic services integrated with other relevant health services, rooted in the primary care level, with proper referral channels to existing genetic centers; prioritize prevention programs and services according to prevalence, severity and predicted outcomes of interventions; train health professionals in genetics; educate the public in genetics; encourage the formation and support of parent/patient organizations; and respect ethical principles and cultural diversity.

World Medical Association Declaration of Helsinki, Ethical Principles for Medical Research Involving Human Subjects, 2000 (http://www.wma.net/e/policy/17-c_e.html)

The World Medical Association Declaration of Helsinki was originally adopted by the 18th World Medical Assembly in 1964 and has subsequently been revised (1975, 1983, 1989, 1996, 2000). The Declaration provides ethical guidance to physicians and other participants in (bio)medical research involving human subjects. 'It is the duty of the physician to promote and safeguard the health of the people. The physician's knowledge and conscience are dedicated to the fulfillment of this duty' (Principle 2).

When medical research is combined with medical care, additional standards apply to protect the patients: 'in the treatment of a patient, when proven prophylactic, diagnostic and therapeutic methods do not exist or have been ineffective, the physician, with informed consent from the patient, must be free to use unproven or new prophylactic, diagnostic and therapeutic measures, if in the physician's judgement it offers hope of saving-life, re-establishing health or alleviating suffering. Where possible, these measures should be made the object of research, designed to evaluate their safety and efficacy' (Principle 32).

Organization for Economic Co-operation and Development, GENETIC TESTING Policy Issues for the New Millennium, Paris, OECD, 2000

An OECD workshop on genetic testing held in Vienna on 23-25 February 2000 was devoted to the discussion of ways

to optimize health care benefits while protecting individuals and their families from the potential of discrimination on the basis of the testing. Participants identified four areas where coordinate international action is urgently needed: (1) Development of internationally recognized and mutually compatible best practice policies for quality assurance and accreditation of genetic tests and services; (2) Development of compatible electronic information systems in genetics; (3) Enhancement of current counseling services, genetic training and public information; and (4) Examination of possible impacts of monopolistic licensing practices.

European Society of Human Genetics, Proposed statement on Formal recognition of medical genetics as a medical specialty in Europe, June 2001 (http://www.eshg.org)

The European Society of Human Genetics recommends a *Formal recognition of medical genetics as a medical specialty in Europe* in order 'to aid the provision and development of genetic services for individuals and families in Europe'. (…) 'The ESHG believes that there are many advantages for the specialty to be recognized internationally, in particular to enable the full impact of the Human Genome Project to be translated into practice across all specialties'. (…) 'The ESHG believes that the benefits of recognizing medical genetics as a specialty will include (1) the establishment and implementation of training programmes; (2) the identification of resources required for service and training; (3) recruitment to the specialty in its own right; (4) the development of relationships between medical geneticists and other specialties; and (5) the dissemination of information to and training for non genetics health professionals'.

World Health Organization, Collaboration in Medical Genetics, Report of a WHO meeting, Toronto, April 2002 (http://www.who.int/ncd/hgn/publications.htm)

Experts recommendations made for WHO included the following:

- To develop and strengthen comprehensive medical genetic services linked to primary health care as the key strategy for the prevention and control of conditions with genetic causation that include genetic counseling, the appropriate use of safe and effective technologies, and the support to parent/patient organizations.
- To assist Member states in establishing undergraduate and postgraduate education programs for the teaching of medical genetics for all health professions (physicians, nurses, psychologists, public health professionals, etc); in developing training modules on genetic counseling and application of genetics/genomics technologies in clinical practice; and in improving awareness of genetics among policy makers, community leaders, patient/parent organizations, journalists, and the general public.
- To assist Member states in assembling regional expert interdisciplinary advisory groups to recommend practical regulatory systems which will ensure the safety and effectiveness of medical applications of new genetic/genomic technologies before they are introduced on the market.

European Institutions *European Union, Council Directive 93/16/EEC of 5 April 1993 to Facilitate the Free Movement of Doctors and the Mutual Recognition of their Diplomas, Certificates and other Evidence of Formal Qualifications* (http://www.ilo.org/public/english/employment/skills/recomm/instr/eu_5.htm)

The EU directive facilitates the free movement of doctors and the mutual recognition of their diplomas, certificates and other evidence of formal qualifications. Article 4 states that each Member State will recognize the formal qualifications in specialized medicine awarded to nationals of Member States by the competent authorities or bodies of other Member States. Article 6 states that some countries award qualifications in a specialized branch of medicine which has been formally constituted by national regulations in that country, but that the branch of medicine may not be formally recognized for all Member States. Article 24 lays down minimum requirements for training leading to a formal qualification in specialized medicine.

The Group of Advisers on the Ethical Implications of Biotechnology to the European Commission, Opinion No. 6 on Ethical Aspects of Prenatal Diagnosis, 1996

The Group of Advisers to the European Commission on the Ethical Implications of Biotechnology considers that 'the offer and use of prenatal diagnosis presuppose good quality social and medical services, especially adequately trained staff, suitable equipment and reliability of the techniques. Safeguards against unethical or unprofessional practices must be in place for all centers offering these procedures. These centers must be officially recognized. Because the consequences of the information can be of the greatest importance to all concerned, it is an ethical imperative that counseling, which requires a specific competence, should be of good quality and widely available. This implies that there must be sufficient trained medical, nursing and other professionals to provide one-to-one counseling when prenatal diagnosis is performed. In accordance with the subsidiary principle, the European Union should stir to achieve a high and comparable level of quality of the training of the professionals, namely concerning the genetic counseling, and of the services provided in different Member States'.

Council of Europe, Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine, 1997 (http://www.coe.fr/fr/txtjur/164fr.htm)

The Council of Europe is at the origin of the first international convention in the field of bioethics. The Convention is the first internationally binding legal text designed to protect people against the misuse of biological

and medical advances. This text has legal effect in the Council of Europe's member States that have ratified it. The Committee of Ministers of the Council of Europe has also taken the issues of predictive medicine in a series of recommendations.

The Convention sets out to preserve human dignity, rights and freedoms, through a series of principles and prohibitions. According to Article 5, a genetic test 'may only be carried out after the person concerned has given free and informed consent to it'; Article 12 states that 'tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counseling'. The restriction of genetic diagnostics to health or scientific purposes is reinforced by Article 11, which states that 'any form of discrimination against a person on grounds of his or her genetic heritage is prohibited'. Article 13 forbids germ-line therapy. An additional protocol on the prohibition of human cloning was added in January 1998.

The Committee of Ministers of the Council of Europe: Recommendations

Before the member States of the Council of Europe, the other States and the European Community signed the Convention for Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine, the Committee of Ministers of the Council of Europe took the issues of medical genetics under consideration in a series of Four recommendations:

- Recommendation No. R (90) 3 on prenatal genetic screening, prenatal genetic diagnosis and associated genetic counseling
- Recommendation No. R (92) 3 on genetic testing and screening for health-care purposes
- Recommendation No. R (94) 11 on screening as a tool of preventive medicine
- Recommendation No. R (97) 5 on the Protection of Medical Data

The principles contained in these recommendations governed (1) the rules for good practice (informing the public, quality of genetic services, criteria for selecting diseases suitable for testing, counseling, economic aspects, quality assurance), (2) access to genetic tests (equality, self-determination, non compulsory nature of tests, non discrimination, privacy), (3) data protection and professional secrecy (data protection, professional secrecy, separate storage of genetic information, unexpected findings), and (4) research (supervision, handling of data).

Council of Europe, Recommendation No. R (90) 3 on Prenatal Genetic Screening, Prenatal Genetic Diagnosis and Associated Genetic Counseling, 1990 (http://www.coe.fr/cm/ta/rec/1990/html)

Governments of Member States are recommended to adopt legislation in conformity with a series of 14 principles or to take other appropriate measures to ensure the implementation of these Principles. Four principles particularly concern the provision of genetic services, Principles 1, 3, 8 and 14. Principle 1 states that 'No prenatal genetic screening and/or prenatal genetic diagnosis tests should be carried out if counseling prior to and after the tests is not available'. Principle 3 stipulates that 'prenatal genetic screening and prenatal genetic diagnosis should only be carried out under the responsibility of a physician; laboratory procedures must be carried out in qualified institutions which have been approved by the state or by a competent authority of the state to conduct such procedures'. According to Principle 8, 'The information given during the counseling prior to prenatal genetic screening and prenatal genetic diagnosis must be adapted to the person's circumstances and be sufficient to reach a fully informed decision. This information should in particular cover the purpose of the tests and their nature as well as any risks which these tests present'. Principle 14 states that 'where there is an increased risk of passing on a serious genetic disorder, access to preconception counseling and, if necessary, premarital and preconception screening and diagnostic services should be readily available and widely known'.

Council of Europe, Recommendation No. R (92) 3 on genetic testing and screening for health-care purposes, 1992 (http://www.coe.fr/cm/ta/rec/1992/92r3.htm)

Governments of Member States are recommended to be guided in their legislation and policy by a series of 13 recommendations to ensure respect for certain principles in the field of genetic testing and screening for health care purposes, including medical research.

Principle 1, 'Informing the public', states that '(a) Plans for the introduction of genetic testing and screening should be brought to the notice of individuals, families and the public; (b) The public should be informed about genetic testing and screening, in particular their availability, purpose and implications – medical, legal, social, and ethical – as well as the centers where they are carried out. Such information should start within the school system and be continued by the media'.

Principle 2, 'Quality of genetic services' states that: '(a) Proper education should be provided regarding human genetics and genetic disorders, particularly for health professionals and the paramedical professions, but also for any other profession concerned. (b) Genetic tests may only be carried out under the responsibility of a duly qualified physician. (c) It is desirable for centers where laboratory tests are performed to be approved by the State or by a competent authority in the State, and to participate in an external quality assurance'.

Principle 3, 'Counseling and support' stipulates that '(a) Any genetic testing and screening procedure should be

accompanied by appropriate counseling, both before and after the procedure. Such counseling must be nondirective. The information to be given (…) must be adapted to the circumstances in which individuals and families receive genetic information'.

Principle 4, 'Equality of access – nondiscrimination' states that: '(a) there should be equality of access to genetic testing, without financial considerations and without preconditions concerning eventual personal choices. (b) No condition should be attached to the acceptance or the undergoing of genetic tests. (c) The sale to the public of tests for diagnosing genetic diseases or a predisposition for such diseases, or for the identification of carriers of such diseases, should only be allowed subject to strict licensing conditions laid down by national legislation'.

Principle 5, 'Self-determination' states that: '(a) the provision of genetic services should be based on respect for the principle of self-determination of the persons concerned. For this reason, any genetic testing, even when offered systematically, should be subject to their express, free and informed consent'.

Council of Europe, Recommendation No. R (94) 11 on Screening as a Tool of Preventive Medicine, 1994 (http://www.coe.fr/cm/ta/rec/1994/94r11.htm)

Governments of Member States are recommended to take account in their national health planning regulations and legislation of the conclusions and recommendations set out in the appendix of this recommendation. 'Because there are differences in health needs and health services, as well as in ethical values and in legal norms and rules between countries, the decision to implement a particular screening program should be taken in cooperation with the medical profession by each country' (Principle 1.7). The organization of a screening program must be tailored to the structures of the preventive and curative systems. 'If appropriate structures in the curative health care system are lacking, screening should not be implemented until they are developed' (Principle 6.3).

Council of Europe, Recommendation No. R (97) 5 on the Protection of Medical Data, 1997 (http://www.coe.fr/data-protection/rec/r(97)5eexp.htm)

Under Chapter 4 on 'Collection and processing of medical data', 'medical data [which includes genetic data] may be collected and processed if permitted by law for preventive medical purposes or for diagnostic or for therapeutic purposes with regard to the data subject or a relative in the genetic line, or to safeguard the vital interests of a data subject or of a third person' (Principle 4.3). Principle 4.4 states that 'if medical data have been collected for preventive medical purposes or for diagnostic or therapeutic purposes with regard to the data subject or a relative in the genetic line, they may also be processed for the management of a medical service operating in the interest of the patient, in cases where the management is provided by the health-care professional who collected the data, or where the data are communicated in accordance with principles 7.2 and 7.3 [on the conditions of communication]'. Regarding genetic data, 'the collection and processing of genetic data should, in principle, only be permitted for health reasons and in particular to avoid any serious prejudice to the health of the data subject or third parties. However, the collection and processing of genetic data in order to predict illness may be allowed for in cases of overriding interest and subject to appropriate safeguards defined by law' (Principle 4.9).

Organisation for Economic Cooperation and Development, Genetic Testing: Policy Issues for the New Millennium, 2000 (http://www1.oecd.org/dsti/sti/s_t/biotech/act/gentest.pdf)

In February 2000, the Organization for Economic co-operation and Development (OECD) held a workshop on 'Genetic Testing: Policy Issues for the New Millennium' in Vienna. The principal goal of the workshop was to consider whether the various approaches of OECD Member countries for dealing with new genetic tests are appropriate and mutually compatible. Participants identified a number of policy areas requiring international coordination and the establishment of coherent international policies.

European Union, Charter of Fundamental Rights of the European Union, 2000

Article 35 on 'Health care' of the Charter states that 'Everyone has the right of access to preventive health care and the right to benefit from medical treatment under the conditions established by national laws and practices. A high level of human health protection shall be ensured in the definition and implementation of all Union policies and activities'.

European Parliament, Temporary Committee on Human Genetics and Other New Technologies in Modern Medicine, Report on the ethical, legal, economic and social implications of human genetics, 2001 (http://www.europarl.eu.int/comparl/tempcom/genetics/rapfin/rapfin_en.doc)

On 13 December 2000 the European Parliament decided to set up a temporary committee on human genetics and other new technologies in modern medicine, which was to remain in existence for 1 year. According to the brief conferred on it, the committee had the tasks of:

- compiling as complete an inventory as possible of new and potential developments in human genetics and of their uses, so as to provide Parliament with a detailed analysis of such developments necessary to enable it to assume its political responsibilities;
- examining the ethical, legal, economic, and social problems posed by such new and potential developments and by their uses;
- examining and recommending to what extent the public interest requires a proactive response to such developments and uses;
- providing an orientation for Parliament and the other Community institutions with regard to research in human genetics and other new technologies.

Council of Europe, Recommendation 1512: Protection of the Human Genome, 2001 (〈http://star.coe.fr/ta/TA01/EREC1512.htm〉)

The Council of Europe's Parliamentary Assembly notes that the human genome international research project, in view of the numerous and unimaginable consequences that it might have for medicine, conjures up scenarios for all humanity that raise numerous ethical questions, while holding out the promise of enormous improvements in the quality of life. The genetic age will dawn with the completion of the project: diagnosis will become objective, and it will be possible to identify the presence of genetic disorders or a genetic predisposition to illnesses at an early stage. In many cases, gene therapy will become possible, and this will basically give rise to a form of genetic engineering designed. At the same time, the Assembly is aware of the enormous ethical implications of further research on the human genome, including some of a negative nature. These include questions regarding the cloning of cells, the conditions ruling genetic testing and the divulging and use of obtained information.

The Assembly calls, *inter alia*, through the establishment of a Euroforum on Human Genetics, for the widest possible participation by citizens in the discussion on the human genome through the involvement of the European media and suitable and accurate information by the Council of Europe.

European Countries The provision of genetic services is not specifically legislated in most European countries. Genetic testing legislation has been implemented in Austria, Belgium, France, Norway, Sweden, Switzerland, and the Netherlands. Denmark, Germany and the United Kingdom have issued policy statements or recommendations on the application of genetic tsting. In addition, most countries have laws regulating the termination of pregnancy and these directly or indirectly regulate also the use of prenatal diagnostics. There are recommendations concerning genetic services provided by different actors including human genetics societies and societies of clinical geneticists in many countries. In the following the laws and also the less formal recommendations whenever known to us are presented for each country.

Austria *The Gene Technology Act (Law BGB 510/1994), 1994* (http://www.gentechnik.gv.at/gentechnik/B1_orientierung/gen_10084.html)

The 'Gene Technology Act' regulates genetic testing. Gene analysis, as it is defined in this Act, comprises molecular biological investigations for the identification of disease-causing mutations. Such examinations are allowed only for research or medical purposes. According to this act, laboratories where genetic tests for the diagnosis of a predisposition or for the identification of a carrier status of inherited diseases are performed have to be accredited by the competent authority. Genetic tests for the diagnosis of manifested diseases do not require an authorization but are subject to strict measures for data protection.

To carry out predictive genetic testing, laboratories have to meet a number of specific requirements. These include quality of the technical equipment, adequate qualification and experience of the performing staff, appropriate confidentiality measures. Genetic counseling has to be carried out before and after genetic testing, and has to include psychological and social considerations as well. The patient has to provide written informed consent prior to the performance of a predictive genetic test.

In addition to the Gene Technology Act, on 23 January 1998, the Austrian Advisory Board on Gene technology (Österreichische Gentechnikkommission) adopted a set of additional criteria and requirements (Kriterienkatalog) for predictive genetic testing. This Kriterienkatalog is not legally binding but gives guidelines to which relevant institutions and the competent authority should adhere. It is available on the government's home page http://www.gentechnik.gv.at under Rechtliches - Gentechnikbuch.

Belgium Belgium was one of the first countries in Europe to form a Council for Human Genetics. Since 1973, the 'De Hoge Raad voor de Antropogenetica – or 'Conseil supérieur de la Génétique humaine' has represented the genetic centers of Belgium in the respective university hospitals. In 1987, the country developed legislation (see below) to restrict genetic counseling and diagnostic testing to these centers. These centers are in general financed by government and are obliged to deliver genetic counseling along with the tests. Genetic services are accessible to everybody who needs them, which means that referral by a physician is not necessary.

Although there exists no formal training program for clinical/medical geneticists in Belgium, a clinical specialization in one of the other medical specialties is strongly advised, together with several years of training in a genetic center, with at least part of the time spent in a molecular and/or cytogenetic laboratory.

Royal Decree of 14 December 1987 concerning the degree of standards, which have be fulfilled by the centers for human heredity

This decree states that genetic diagnostic testing could only be carried out in the recognized laboratories of the genetic centers. Each recognized genetics center, which performs genetic tests in the accredited laboratory, should in conjunction with the laboratory activities offer clinical diagnostic and genetic counseling services. In addition, each of these centers must provide a detailed activity report on yearly basis for the government. On this condition, the genetic centers receive funding from the government. The 1987 legislation also says that genetic counseling should be offered on a nonprofit, multidisciplinary basis and includes

all necessary psychological and moral support to help the individual deal with the information and the implications.

Cyprus Cyprus has no specific legislation dealing with human genetics yet, and preparatory work in this area is in its early stages. However, Cyprus has subscribed the European Protocol for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine.

Clinical genetic services are provided by public and private centers. Laboratories do not need special accreditation or license to practice in Cyprus and no system for accreditation or licensing has so far been established. Laboratories take part in external quality assessment on an individual basis. There are no formal training programs in genetics by Cypriot academic institutions. Human reproduction techniques and PGD are provided in Cyprus but there is no law to regulate practice.

Czech Republic Czech Republic has no specific legislation dealing with human genetics yet. However, Czech Republic has subscribed the European Protocol for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine.

Clinical genetics has been officially included in the health-care system since 1980. There is a strong demand for individual laboratories and departments' accreditation. Systematic postgraduate education in clinical genetics has expended since 1980.

Denmark In Denmark genetic testing is mainly regulated through the legal frameworks that apply to the Danish national health-care system as a whole (see below). In addition, some specific guidelines have been developed, such as the following:

- *Danish National Board of Health, Guidelines and recommendations for indications for prenatal diagnosis (1994)*
- *Danish Ministry of Health, Guidelines for the Information of Relatives in HNPCC testing (1996)*
- *Danish Council of Ethics, Priority-setting in the Health Service (1997)*
- *Danish Council of Ethics, Report and Recommendations on Presymptomatic Genetic Testing (2000)*
- *Danish HNPCC Registry, Guidelines for Counseling Testing and Follow-Up Programs for NHPPC*
- *Danish Breast Cancer Collaboration Group, Guidelines for Counseling Testing and Follow-Up Programs for BRCA 1 and 2*

Clinical genetics became a medical specialty in 1996 and genetic counseling is performed by specialists in clinical genetics. DNA testing is performed in clinical genetic and clinical biochemistry departments, mainly in university hospitals. Laboratories do not need special accreditation or license to practice and no system for accreditation or licensing has so far been established. However, laboratories take part in external quality assessment on an individual basis.

Estonia *Act No. 1-5/829/1996 on Newborn Screening, Social Ministry, 1996*

The organization, performance and availability of newborn screening for phenylketonuria and hypothyreosis are coordinated by this Act.

Regulation No. 33/1997 on Prenatal Diagnostics, Social Ministry, 1997

The performance of prenatal testing, availability and quality control are regulated in this document.

Finland Genetic testing is carried out in university hospitals and in specialized private laboratories. Although no specific regulations exist on genetic testing, supervision and quality control of both public and private sector laboratories are organized by state authorities. A general quality assessment scheme of genetic testing has so far not been developed. However, a recent Working Party set up by the Ministry of Social Affairs and Health has made recommendations concerning quality assessment, supervision, counseling and use of information in relation to genetic testing. The Ministry will decide on possible legislative measures. There is also an Advisory National Ethics Board, since 1998, which can discuss matters in the field of genetic services.

Act on the Status and Rights of patients, 785/1992

The act regulates that is patient's right to be informed about his/her state of health, patient's right to self-determination, drafting and keeping patient documents and confidentiality of information in patient documents. Following the publication of this Act a National Advisory Board on Health Care Ethics (1998) was formed which takes initiatives and releases statements and recommendations on ethical issues in health care.

Act concerning health care professionals, 559/1994

The aim of the act is to promote the safety of patients and to improve the quality of health-care services by ensuring that health-care professionals have the necessary training and professional qualifications and by organizing the supervision of health-care professionals.

Gene Technology Act, 1995

This act aims to promote the safe use and development of gene technology in an ethically acceptable way, and to prevent and avert any harm to human health. It does, however, not apply to modification of human genetic material by genetic techniques. An amendment of this Act is in preparation.

Medical Research Act, 2000

This act includes research using human embryos, up to the age of 14 days postconception, by a specific permission from a statutory board. Preimplantation genetic diagnosis is an accepted field of study.

France *Laws No. 94-653 of July 29, 1994 on respect for the human body* (http://www.cnrs.fr/SDV/loirespectcorps.html)

This law modifies the Civil Code by introducing notably the notions of the fundamental right to respect for one's body, therapeutic necessity as the only acceptable reason for violating bodily integrity and this only if the individual has consented. Chapter III of the law is devoted to 'Genetic characteristics and genetic identification of a person'. Article 16-10 states that the genetic study of a person's characteristics may only be undertaken for medical or scientific research purposes. Before such a study is undertaken the person's consent must be secured. Strict penalties are provided if consent has not been obtained (Article 226-25 of the Penal Code) or if the genetic study is carried out for non medical or non scientific purposes (Article 226-26 of the Penal Code). The restriction of genetic testing for medical or scientific purposes has been reaffirmed in the Article L' 145-15 of the new title VI of the Public Health Code (1998).

Laws No. 94-654 of July 29, 1994 on the donation and use of elements and products of the human body, medically assisted procreation and prenatal diagnosis (http://www.cnrs.fr/SDV/loirespectcorps.html)

Prenatal diagnosis must be preceded by a medical genetic counseling consultation. The cytogenetic and biological analyses must be carried out in authorized establishments. Preimplantation diagnosis is only allowed in certain circumstances.

National Ethical Consultative Committee for the Life and Health Sciences in France, Genetics and Medicine: From Prediction to Prevention, Paris, 1995 (http://www.ccne-ethique.org/english/avis/)

This report declares the ethical principles that must be respected, with respect to all the activities involved in genetics and medicine. Its recommendations cover the following topics and ethical principles: respect of the autonomy of the subject, respect of medical confidentiality; respect of privacy in computerizing personal data; the use of biological samples; the prohibition of using results of genetic tests for purposes other than medical or scientific; procedures of accreditation of the materials involved in genetic testing; prior evaluation of the impact of the tests; information and formation of all medical personnel in genetics; the need to guarantee correct public information; prohibition of all uses that would contribute to stigmatization or unfair discrimination in the social and economic spheres.

National Consultative Ethics Committee, Review of the Law No. 94-653 of July 29, 1994: propositions regarding preimplantation diagnosis and prenatal diagnosis, 1998 (http://www.ccne-ethique.org/english/avis/)

Concerning preimplantation diagnosis, the Article L. 162-17 of the law, paragraph 5 states that biological diagnosis using cells taken from an in-vitro embryo is only authorized in exceptional circumstances. The National Consultative Ethics Committee recognizes the exceptional nature of this diagnosis, which concerns incurable diseases of particular severity, which have been identified in one parent. However, it notes the apparent contradiction in the text of the law between reference to the incurable nature of a disorder and the possibility of treating it.

According the Article L. 162-16, paragraph 1 of the law, prenatal diagnosis refers to medical practices with the aim of detecting in utero in an embryo or fetus a particularly severe disorder. There must be a prior medical genetic counseling session. The National consultative Ethics Committee recommends that it should be mentioned that the obligation to conduct a prior medical counseling session, as stipulated in the text of the law, only refers to the biological diagnosis.

Decree no. 2000-570 dated June 23, 2000 fixing the conditions of prescription and implementation of genetic characteristics and genetic identification investigations of a person for medical reasons and modifying the Public Health Code

This decree delineates 5 conditions for prescribing and implementing genetic testing for medical purposes: (1) Condition of prescription; (2) Condition of approval from appropriate authorities both for clinicians and laboratories; (3) Conditions of reporting results; (4) Conditions of medical record protection; and (5) Approval from the National Consultative Commission created for this purpose.

Physicians responsible for this genetic analysis must be qualified in medical biology or biology-pharmacology. Exceptionally, a senior scientist (non-MD) may be responsible for these genetic analyses only if he/she has experience on cytogenetics or molecular biology. A consultative Commission must be asked to rule on the necessity of such procedures and on their implementation.

National Consultative Ethics Committee, Consent for the benefit of another person, 2001 (http://www.ccne-ethique.org/english/start.htm)

In this opinion, the National Consultative Ethics Committee opposes the legal and the ethical considerations on this topic. It considers that consent in favour of, or for the benefit of a third party, leads to several principles, possibly conflicting, being considered: the autonomy of the index person, benevolence in favour of a third party, and solidarity. In the last analysis, the committe consider that educating society to a better understanding of the meaning of solidarity, is a means of respecting individuals by calling on their sense of responsibility, and informing them on the purpose and altruism of a decision. To consent in the interest of another person is to be both separate and responsible.

Law no. 2002-303 of March 4, 2002 relating to the rights of the patients and the quality of the health care system (http://www.assembleenat.fr/dossiers/droits_des_malades.asp)

The goals of this act is:

- To develop the medical democracy (first title) by recognizing rights for any person in its relationships with the health-care system, by granting rights to the users and by associating them to the operation of the health care system, and by allowing the development of policies of health at the national and regional levels;
- To improve quality of the health-care system (title II) by developing competences of the professionals, the continuous medical training and a global prevention policy;
- To allow the repair of the medical risks (title III) by improving the insurance access, by defining the principles of the medical responsibility and by creating procedures for amicable agreement and for the compensation of the medical accident victims.

Germany As regards the application of genetic testing, professional organizations and vocational associations have issued a large number of comments and guidelines (see below). These comments and guidelines are based on the principles of counseling and education, autonomy and confidentiality. However, they do not have a legally binding character, but are only recommendations to their members.

Attempts are being made by some institutions to review and discuss the technical possibilities of PGD, the question of its necessity, and the ethical, social, and legal problems, in particular the necessary changes of the Embryo Protection Law and the professional guidelines for IVF. In addition, the reformed Abortion Law of 1995 still bans abortion but allows exceptions under certain conditions. The jurisdiction in the German constitution protects the diseased or disabled. Therefore a future disorder or disability of the fetus cannot be used as sole reason for abortion.

The German Bundestag, Chancen und Risken der Gentechnologie Enquete-Commission, 1987

Prenatal diagnosis and newborn screening programs were accepted. The report contained detailed recommendations on the consent and counseling requirements, which must be fulfilled before any genetic test can be carried out. In most instances the report did not recommend that legislation be enacted but rather that these matters be supervised by authoritative professional bodies.

The German Bundestag, The Embryo Protection Law, 1990

This law regulates medical actions around *in-vitro* reproduction. IVF is restricted to cases of infertility.

The Board of Medical Genetics, Statement on carrier screening for cystic fibrosis, 1990

The German Society of Human Genetics, Statement on prenatal diagnosis of sex, 1990 (http://gfhev.de/kommission/index.html)

'The German Society of Human Genetics considers the use of prenatal diagnosis to choose infant sex as indefensible. To prevent misuse of information about the infant's sex, (…) the parents and obstetricians should not be informed of the child's sex chromosome constitution until after the end of the 14th week of pregnancy. The only exceptions are for sex chromosome disorders and sex-linked hereditary disorders. If prenatal diagnosis from a trophoblast biopsy is indicated and the parents agree to the procedure only if the sex of the child is made known to them with the chromosomal findings before the end of the 14th week of pregnancy, the biopsy and diagnosis should be refused and the parents should be referred for an amniocentesis with subsequent amniotic fluid cell analysis'.

The German Society of Human Genetics, Statement on postnatal predictive genetic testing, 1991 (http://gfhev.de/kommission/index.html)

Predictive genetic testing must take, among other things, the following into consideration: (1) 'Comprehensive information must be offered to all concerned persons, and counseling about alternative options must be guaranteed'. (…) (3) 'Explanation and counseling about available tests must be non directive. (4) Predictive genetic diagnosis may be performed only for persons of legal age. Exceptions are for disorders for which preventive or therapeutic measures could be initiated in childhood'. (…) (6) 'Predictive genetic diagnosis must not become a routine investigation. When developing guidelines, the expectations of the affected should be extensively considered as was done internationally. (…) Since manifold problems are foreseeable, predictive genetic diagnosis should be introduced only within the framework of a scientifically accompanying pilot project. Due to their limited personnel and equipment and in spite of professional competence, human genetics institutes and genetic counseling facilities presently are able in only a limited way to guarantee that predictive genetic diagnosis is carried out within the required framework. However, attempts should be made to establish this type of diagnosis including the required counseling, at qualified non profit institutions'.

The German Society of Human Genetics, Statement on carrier screening, 1991 (http://gfhev.de/kommission/index.html)

The Board of Medical Genetics, The German Society of Human Genetics, Moratorium on maternal serum markers screening, 1992

The German Society of Human Genetics, Curriculum for non-MD human geneticists, 1993, 1994 (http://gfhev.de/kommission/index.html)

The Board of Medical Genetics, Patient information, Informed consent for genetic counseling, 1994

The Board of Medical Genetics, Statement and Recommendation on confidentiality, 1995

The German Society of Human Genetics, Statement on BRCA1 testing, 1995 (http://gfhev.de/kommission/index.html)

The framework required for BRCA1 gene testing must take, among others, the following principles into

consideration: 'a guarantee that comprehensive information will be offered, including counselling about alternative approaches; (…) introduction within the framework of scientifically accompanied pilot projects; inclusion of competent medical geneticists and genetic counselors'.

The German Society of Human Genetics, Statement on genetic diagnosis in childhood and adolescence, 1995 (http://gfhev.de/kommission/index.html)

'Genetic diagnosis in children and adolescents is indicated if it is necessary for the differential diagnosis of manifest symptoms or for establishing the etiology of a disease. A predictive genetic diagnosis is indicated during childhood if the onset of a disorder can be regularly expected at this age and if medical measures can be taken to prevent the disease or its complications or to treat the disease. (…) However, deferring a predictive genetic diagnostic test should not prevent discussing the disease in question with the child in a manner appropriate to his/her age, including how it is inherited and the possibility of its being diagnosed. (…) An investigation for the sole purpose of determining the carrier status for a recessive inherited illness or a balanced familial chromosomal translocation should not be carried out since the results would only be significant for future reproductive decisions of the child him/herself. Therefore the examination should be deferred until the child can understand all the associated facts and psychosocial implications and asks for the test him/herself'.

The Board of Medical Genetics, The German Society of Human Genetics, Statement on new abortion law, 1995

The German Society of Human Genetics, Statement on preimplantation diagnosis, 1995 (http://gfhev.de/kommission/index.html)

'The German Society of Human Genetics is of the opinion that preimplantation diagnosis, that is legal within the framework of professional regulations, should basically be made available to all women who carry a specific genetic risk for a severe infantile disease or developmental disorder and who would like to have the risk clarified by this method. Because of the inherent problems of preimplantation diagnosis, the framework for the procedure must meet high requirements'.

The German Society of Human Genetics, Position Paper, 1996 (http://gfhev.de/kommission/index.html)

This paper defines standards for the application of genetic tests to nearly all fields of practical genetics. Concerning access and use of genetic services, 'all population groups should have similar access to genetic information, counseling, and diagnostic services. Information should be generally available, appropriate, and qualified, and counseling and examination capacities must be adequate.

Because of the impact of genetic diagnoses, utilization of genetic counseling and diagnosis should occur on a voluntary basis only. (…) Thus, everyone has the right not to know about his or her own genetic make-up. Likewise, no one should be prevented from using genetic counseling and diagnostic services. Individuals who utilize certain genetic examinations, but also persons who refuse to utilize them are in danger of being stigmatized or discriminated against. Such tendencies of public opinion must be counteracted by increased efforts to inform and educate the public.

(…) At this time, the only known exception to the principle that the utilization of diagnostic genetic tests be voluntary is the routine examination of newborns for genetically determined disorders that are amenable to early treatment or prevention'.

The Board of Medical Genetics, The German Society of Human Genetics, Declaration, Curriculum on Education in ethical and psychological dimensions of genetic counseling, 1996

The Board of Medical Genetics, Guidelines on genetic counseling, 1996

The Board of Medical Genetics, Guidelines on tumor cytogenetic testing, 1996

The Board of Medical Genetics, Guidelines on molecular genetic testing, 1996

The Board of Medical Genetics, Guidelines on cytogenetic testing, 1997

The Board of Medical Genetics, Guidelines on molecular genetic testing of CVS material, 1997

The Board of Medical Genetics, Guidelines on molecular genetic testing for cystic fibrosis, 1997

The Board of Medical Genetics, Guidelines on molecular genetic testing for Fra-X, 1998

The German Society of Human Genetics, Guidelines on aneuploidy testing of uncultivated AC, 1998 (http://gfhev.de/kommission/index.html)

The German Medical Association, Guidelines on predictive genetic testing for tumor disposition, 1998

The Board of Medical Genetics, Guidelines on molecular genetic testing for DMD/BMD, 1999

The German Medical Association, Guidelines on predictive genetic testing, in preparation.

Greece Although the first law on the regulation of the practice of medical genetics was passed by the Greek Parliament in 1980, it was never implemented. A special advisory committee was formed in the Central Health Council of the Ministry of Health and its proposals for the development of genetics centers and the specialty of genetics are being studied by the Ministry in order to be incorporated in a forthcoming Health Bill.

Medical genetics has entered the university curriculum as an integral part of medical and nursing studies, through the establishment of a department of genetics in the medical school and the teaching of the medical genetics and genetic counseling at the undergraduate, graduate and postgraduate level in both medical and nursing faculties respectively.

Quality control for cytogenetic and molecular studies is performed by several university and private genetic laboratories. At present, genetic counseling is performed at certain university and public genetic units by clinical geneticists or privately, mostly for specifically referred cases.

The proposal to the Ministry of Health is for the development of genetic units in all university and district hospitals according to the Council of Europe guidelines.

Hungary There are no approved guidelines for genetic testing in Hungary. Professionals in university or municipal hospitals are delivering services according to practice based on medical literature, nation-wide and international experience in genetic counseling and discussions at scientific meetings. In 1999, an Ad Hoc Committee was named by the Ministry of Health to develop guidelines for genetic screening and testing in Hungary.

No agency has jurisdiction over clearing diagnostic services for marketing. However, there is occasional collaboration between service delivery units and industry which supplies kits for which licensing has been obtained.

Iceland Iceland has no law that specifically deals with human genetics.

Act n. 97/1990 on a Healthcare Services, Ministry of Health, 1990 (http://brunnur.stjr.is/interpro/htr/htr.nsf)

The health sector is regulated according to this Health Service Act by which all inhabitants have right of access to the best possible health service at any given time for the protection of their mental, social and physical health.

Act n. 74/1997 on the Rights of Patients, Ministry of Health, 1997 (http://brunnur.stjr.is/interpro/htr/htr.nsf)

This Act includes fundamental rights of patients including rules on consent, confidentiality and handling of information in clinical records.

Act n. 139/1998 on a Health Sector Database, Ministry of Health, 1998 (http://brunnur.stjr.is/interpro/htr/htr.nsf)

This Act is in compliance with the Act on the Rights of Patients. By reference to article 29 in the Act on the Rights of Patients, the Minister of Health and Social Security has issued a regulation on scientific research in the health sector (Reg. No. 552/1999).

The Act on a Health Sector Database makes it legal for a private company to construct an electronic database of nonpersonally identifiable health data with the aim of increasing knowledge in order to improve health and health services. The Act makes it possible to combine and analyze health data with genetic and genealogical data.

Ireland Ireland has no law specifically dealing with human genetics and Ireland has not signed the 1997 Oviedo Bioethics Convention. Clinical Genetics is a specialty recognized by the Irish Medical Council, and clinical practice is subject to General Medical Council guidelines. A Department of Health committee is currently considering guidelines for assisted reproductive practice, including preimplantation genetic diagnosis.

Ireland has been involved with the UK (Clinical Molecular Genetics Society) and the Netherlands in developing laboratory guidelines for molecular genetic testing for specific diseases. These guidelines have been adopted by the European Molecular Genetics Quality Network (EMQN) (http://www.emqn.org).

Italy *The Italian Committee on Bioethics, Gene Therapy, 15 February, 1991*

The Italian Committee on Bioethics, Prenatal Diagnosis, 17 July 1992

The Italian Committee on Bioethics, The human Genome Project, 18 March, 1994

National Council of the Federation of the Colleges of Physicians and Dentists, the new Italian code of medical ethics, 1995

In this code, article 42 address interventions on genome and conceptuses.

The Italian Committee on Bioethics, Identity and Rights of the Embryo, 22 June 1996

The Italian Committee on Bioethics, The anencephalic newborn and organ donation, 21 June, 1996

National Guidelines for Genetic Testing, 1998

In 1998, National Guidelines for Genetic Testing were prepared by a Task Force appointed by the National Committee for Biosecurity and Biotechnologies, coordinated by the National Health Institute. The general objectives are: (1) ensuring the safety and effectiveness of both existing and newly introduced genetic tests; (2) defining the criteria for quality assurance of laboratories performing genetic tests; (3) ensuring both adequate counseling and the free decision of individuals and families; this will include a particular attention to problems concerning ethics and privacy. Some topics deserving a specific concern have been identified, namely: genetic testing for prenatal diagnosis, genetic testing for susceptibility to cancer and genetic testing for rare diseases. There is no law for preimplantation genetic diagnosis.

Cytogenetic and Molecular Testing in Italy, ISTISAN Reports no.20, 1998

Decreto Presidenziale, 9 luglio 1999 (*Gazzetta ufficiale 22/7/99), Accertamenti per la diagnosi delle malformazioni* (*Art. 1)*

This decree addresses the screening of the following diseases: cystic fibrosis, phenylketonuria and congenital hypothyroidism. It establishes that these services must be free of charge.

The Italian Committee on Bioethics, Orientamenti bioetici per i test genetici, 19 November 1999 (http://www.palazzochigi.it/bioetica/orientamenti%20biomedici.htm)

Lithuania *Act No. 136/1991, Ministry of Health*

In 1991 a University Hospital Human Genetics Center (Vilnius) was created. The activities of the Center focus on

the prevention of inherited diseases, including genetic counseling, neonatal screening for PKU and for congenital hypothyroidism, registration of congenital anomalies and prenatal diagnosis as well as education in human and clinical genetics for medical students. Residentship in clinical genetics was introduced in 1992. The Center takes part in external quality assessment of newborn screening for PKU and congenital hypothyroidism since 1996, Huntington's disease since 1998, and Duchenne muscula dystrophy and cystic fibrosis since 2000.

Act No. 199/1991, Ministry of Health

Clinical genetics became a medical specialty and genetic counseling is performed by specialists in clinical genetics (MD).

Act No. 74/1992, Ministry of Health

This Act regulates the registration of congenital anomalies. Any medical doctor who has diagnosed congenital anomalies at birth must notify it to the Lithuanians Registry of congenital Anomalies at the Vilnius University Human Genetics Center.

Act No 706/1997, Ministry of Health

This Act regulates national standards for genetic counseling and professional responsibilities of clinical geneticists.

Act No. 354/2000, Ministry of Health

The main activities of the Vilnius University Hospital Human Genetics Center are being performed according to the program 'The structure, defects, and protection of gene pool of the Lithuanian population'.

Act No. VIII-1679/2000, Lithuanian Parliament

This law on Bioethics regulates genetic testing. Genetic testing can only be carried out for medical or scientific purposes and only after written consent has been obtained from the individual.

Norway *Act Relating to the Application of Biotechnology in Medicine, Law n. 56 of 5 August 1994* (http://www.helsetil-synet.no/htil/avd2/bio_act.htm)

This Act gives a frame of general guidelines for assisted reproductive technology applications, research on embryos, preimplantation diagnosis, prenatal diagnosis, genetic testing after birth and gene therapy. This Act also specifies obligations about authorization of institutions applying medical biotechnology and the duty for such institutions to report regularly on their activities to the Ministry of Health and Social Affairs.

Genetic testing for diagnostic purposes is permitted without restrictions, but the law requires that comprehensive genetic counseling be given before, during and after genetic tests performed on healthy persons for presymptomatic, predictive or carrier purposes. Presymptomatic, predictive and carrier testing is limited to individuals above the age of 16 years. When the information refers to a diagnostic test, genetic results may be communicated, without restrictions, between medical institutions authorized to apply medical biotechnology. However, the exchange of genetic information about presymptomatic, predictive or carrier tests is restricted. The Act states that it is prohibited to ask whether a presymptomatic, predictive or carrier test has been performed. Gene therapy is only allowed as somatic cell therapy and individuals below the age of 16 years need the consent of their parents or guardians.

Portugal The Ratification of the 'Convention for the Protection of Human Rights and Dignity of the Human Being and the additional protocol on the prohibition of cloning human beings' was published in January 2001. Some guidelines prepared by a task force were also published by the Ministry of Health. These guidelines are concerned with the ethical and professional rules on genetic testing and prenatal diagnosis namely confidentiality, genetic counseling and genetic testing of children. Genetic counseling before testing late onset diseases (Machado - Joseph and Familial Amyloid Polineuropathy) is usually offered but not yet on recessive carriers familial testing and oncologic diseases. The specialty of medical genetics has been formally recognized in 2000.

Quality assessment schemes for laboratory genetic services are not obligatory. Since 1994, Portuguese laboratories have participated in the European EQA of the European Molecular Genetics Quality Network for cystic fibrosis, Friedensreich's ataxia, Huntington disease and Duchenne muscular dystrophy.

There is no legislation for preimplantation diagnosis.

Act No. 90/97 related to the Abortion

Despacho Ministerial No. 9108/97, Guidelines for Molecular Genetic Diagnosis

Despachos Ministerials No. 5411/97 e No 10325/99, Principles and Practice for prenatal diagnosis

Portaria No. 189/98 related to the Abortion

Convention for the Protection of Human Rights and Dignity of the Human Being and the additional protocol on the prohibition of cloning human beings, 2001

Russia *Ministry of Health, On Further Development of Medical Genetic Services in the Russian Federation, Circular n. 316, December 12, 1993.*

All medical genetic services in Russia are mandated by the principal circular n. 316 issued by the ministry of Health on December 12, 1993 'On Further Development of Medical Genetic Services in the Russian Federation'. Revised and updated version of this circular has been prepared. Regulations govern diagnosis at each medical genetic level, the interrelationships between different levels, and the type of diagnostic procedures and basic equipment. Genetic counseling and prenatal diagnostics services are basic subjects of these circulars. There are no officially approved guidelines for predictive genetic testing.

Genetics is recognized as a medical specialty. Basic education in medical genetics is provided in all medical schools and also in medical faculties of many universities.

Spain There are no approved guidelines for genetic testing in Spain. Consent to undergo any medical tests is granted through General Health Law of 25 April 1986. The Organic Law regulating the automated processing and protection of personal data of 13 December 1999 provides special measures of protection for personal health data.

Quality assessment schemes for genetic services have been addressed in specific areas. In 1996 standard criteria for quality control of cytogenetic and prenatal diagnosis laboratories were issued and currently there are plans to develop quality standards for clinical and molecular genetic services.

In 1999, Spain subscribed and joined the European Agreement for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine.

General Health Law of 25 April 1986

The Royal Decree of 21 November of 1986

This Decree rules out the conditions for the Centers to be authorized to perform therapeutic abortion and prenatal tests, as well the requisites to be filled in by practitioners concerned.

The Act 35/1988 of 22 November on Techniques of Assisted Reproduction, 1988

This law regulates the human reproduction techniques when they are performed by a specialist in authorized public or private medical centers. Article 12 regulates preimplantation and prenatal diagnosis. Articles 14 to 17 permit investigation and experimentation for the treatment and prevention of genetic disorders under determined conditions. Article 159 permits that manipulation of human genes only when the intention is the elimination or the improvement of a serious illness.

The Organic Law regulating the automated processing of personal data of 29 October 1992

The Organic Law regulating the automated processing of personal data of 29 October 1992 provides special measures of protection for personal health data (articles 7.3 and 8).

Guidelines for prenatal cytogenetics, 1996

The Organic Law regulating the automated processing and protection of personal data of 13 December 1999

This law includes automated data and any type of personal data.

Sweden *National Board of Health and Social Welfare, Neonatal screening for metabolic diseases, SOSFS, 1988*

Law 114 of March 1991 on the Use of Certain Gene Technologies within the Context of General Medical Examinations (1993)

This law examines the use of certain genetic technology in medical examinations. There must be permission from the National Board of Health and Welfare. Authorization from this body is required before DNA testing can be carried out. This requirement extends to the use of genetic techniques for diagnostic purposes.

Swedish Society for Medical Genetics, 1994

The Swedish Society for Medical Genetics has brought forward a quality assessment document for clinical genetic units including guidelines for cytogenetic and molecular routines as well as for genetic counseling. This document has been adopted by all the university clinical genetic departments as a minimum standard for quality.

The Ministry of Health and Social Affairs, Guidelines on the use of prenatal diagnosis and preimplantation diagnosis, 1995

These guidelines regulate prenatal diagnoses and include prenatal diagnosis by genetic tests. All pregnant women must be informed about prenatal diagnosis. Screening is in principle to be avoided in connection with prenatal diagnosis. Preimplantation diagnosis may only be used for the diagnosis of serious, progressive, hereditary diseases, which lead to premature death and for which there is no cure or treatment.

National Board of Health and Social Welfare, Genetics and Genetechnology in Health Care. State-of-the-Art and Guidelines for Ethical Considerations, 1999

Switzerland *The Swiss Federal Constitution, 1992*

The Constitution provides laws on human genetic practice and medical-assisted procreation. Article 119 (introduced in 1992 as article 24novies, old numbering) paragraph 2 states that the genetic make-up of an individual may be investigated, registered or divulged only with his/her consent or on the basis of a legal prescription.

The Swiss Academy of Medical Sciences, Medical-ethical Guidelines for Genetic Investigations in Humans, Approved by the Senate of the Swiss Academy of Medical Sciences on 3rd June 1993 (http://www.samw.ch/e/richtlinien/richtlinien_fs.html)

The Swiss Academy of Medical Sciences guidelines are not legally binding, unless cantonal legislation gives them binding force. According to the guidelines, genetic investigations are ethically justified if they serve the following purposes: determination of a predisposition for a hereditary disease or handicap, with a view to appropriate planning for the life of the individual, and family planning; or detection of a predisposition for a particular disease when symptoms have not yet appeared, if effective measures can be taken to alleviate and prevent severe effects of the disease or if the result of the investigation is of immediate relevance for planning for the life of the individual or for family planning.

Genetic investigations must be accompanied by appropriate, nondirective counseling before, during and after the investigation. The decision to carry out, continue or stop the investigation rests exclusively with the patient, who will also decide whether and to what extent he wishes to be

informed of, and to draw conclusions from, the result of the investigation. The voluntary nature of participation in the investigation and the right not to be informed of the result must also be guaranteed.

Act on Procreative Medicine, 1998

Article 5 § 3 stipulates that preimplantation diagnosis is forbidden in either clinical or research settings.

The Netherlands In the Netherlands, genetic services are incorporated in the health care and funded in such away that equal access is guaranteed. The quality of the genetic services is ensured by legislation requiring a license from the government (only 8 centers are licensed and funded by the health insurers). Also the close organizational contact of clinical genetics with research groups of human genetics in medical faculty enable a timely update / introduction of new diagnostic technologies.

As far as legislation is concerned there are regulatory frameworks for the licensing of clinical genetics centers as well as the limitation of unlimited growth of activities and commercial testing. Since a few years there is a law protecting individuals against the request of genetic testing (or information) by third parties and recently and recently, a document on the application of genetics in health care has been published by the Dutch Ministry of Health. This document comments then future organization of genetic services and predictive DNA testing and on various psychosocial and ethical issues related to screening, family counseling and presymptomatic DNA testing. Most of the government's views are in accordance with recommendations by the Dutch Health Council in its advice 'DNA diagnostics in health care' (May 1998).

The Health Council of the Netherlands, Report: Heredity, Science and Society: On the possibility and Limits of Genetic Testing and Gene Therapy, The Hague, 1989

The Council takes a strong position on autonomy, suggesting that every individual owns his or her genetic material and therefore informed consent is necessary for any use of it. However, the physician-patient relationship is regarded as one in which the physician's role cannot be specified entirely in terms of satisfying the interests of the patient. The physician has his or her own responsibilities (e.g., to other parties), which lead to a potential conflict between beneficence and autonomy. The council is of the view that unauthorized disclosure may be permissible under limited circumstances when serious harm can be avoided and has noted that relatives' right to privacy should be a consideration when deciding whether or not a disclosure should be made.

The Health Council of the Netherlands: Committee Genetic Screening, Genetic Screening, The Hague, 1994

This committee has listed criteria which must be met by genetic screening programs prior their implementation. The Dutch Health Council defines genetic screening as 'any kind of test performed for the systematic early detection or exclusion of a hereditary disease, the predisposition to such a disease or to determine whether a person carries a predisposition which may produce a hereditary disease in offspring'. The Council states that 'the program for the early detection and treatment of diseases should involve an important health problem'. However, according to the Council, 'it is up to the individual and parents to determine whether a condition is serious enough to enter a screening program'; genetic screening aims 'to enable people to achieve greater autonomy and to decide upon a course of action that is acceptable to them. Voluntary participation based on well-understood information is an absolute requirement and there must be safeguards for free individuals choice during the whole screening process'. Counseling is also considered important.

The Population Screening Act, 1992 (1996)

This act states that screening by means of ionizing radiation, screening for cancer and screening for serious disorders for which there is no treatment are not allowed without ministerial approval, based on the advice and assessment of the Health Council. A license may be refused if the screening program is scientifically unsound, if it conflicts with statutory regulations or if the risks are found to outweigh any benefits.

The Health Council of the Netherlands, Advisory Report on Gene Therapy, 1997

The opportunities and problems surrounding gene therapy are the subject this report.

The Health Council of the Netherlands, Advisory Report on DNA Diagnostics in Health Care, Publication N. 1998/11 1998

Genetic research provides new opportunities for predicting the occurrence of disease, which were discussed in this report.

The Health Council of the Netherlands, Advisory Report on Clinical Genetic Testing and Counseling, Publication N. 1999/07, 1999

According to this report, regulations on clinical genetic testing and counseling in the Netherlands apply to 'postnatal and prenatal chromosome, biochemical and DNA testing, the clinical removal of fetal material, advanced ultrasound scanning for fetal abnormalities and complex genetic counseling'. The regulations are designed to assure the quality and continuity of the procedures in question, which are regarded as a form of medical care.

The report makes the following recommendations: (1) Genetic counseling and the associated test activities should continue to be concentrated in the nominated centers. (2) The professional groups involved in clinical genetics should have responsibility for drafting and updating quality requirements; in this context, the government's role should be supervisory. (3) Forecasts of the level of provision required in this field should take account of the rapid increase in demand for counseling for hereditary forms of cancer. (4) In addition to the Standing Committee on Genetics, several professional organizations are

involved in developing best practice guidelines, including the clinical genetics centers. The centers' activities are regulated by a single package of legislation (Section 2 of the Special Medical Treatments Act).

Turkey Genetic testing is undertaken by molecular genetic units mostly in university hospitals and recently in a limited number of private laboratories. The formulation of the genetic screening programs by the Ministry of Health and Social Affairs is very new and therefore the regulations for each screening program is going to be performed day by day. However, some of the genetic screening programs are being performed individually by the genetic diagnosis centers.

The Regulation of the Genetic Diagnosis Centers (1998, No. 23368) (Genetik Tani Merkezleri Yönetmeligi)

This regulation is about the best practice standards of the centers of both the public sector and the private laboratories. The quality control and standardization of analysis are not included in this regulation. These are controlled by the Turkish Association of Medical Genetics Committee.

United Kingdom An Advisory Committee on Genetic Testing (ACGT) was established in 1996. Its role was to advise UK Health Ministers on developments in genetic testing, on the ethical, social, and scientific aspects of testing, and on the requirements to be met by suppliers of genetic testing services. It also considered the use, or potential use, of tests both for clinical practice and for those supplied directly to the public. ACGT has published two reports which are relevant to the provision of genetic services: (1) a code of practice and guidance on genetic testing services supplied direct to the public (1997), and (2) a report on genetic testing for late-onset disorders (1998) (see below). The work of the ACGT has now been transferred to the Human Genetics Commission.

Other government and nongovernment advisory groups have also discussed the current organization and commissioning of genetic services, and options for the future. They are presented below.

House of Commons Select Committee on Science and Technology, human Genetics: the Science and Its Consequences, Third Report, HMSO, 1995 (http://www.parliament.the-stationery-office.co.uk/pa/cm199899/cmselect/cmsctech/489/48902.htm)

This report examines the ethical issues arising from genetic technology and recommends the setting up of a Human Genetics Commission to regulate the advance of genetic technology.

The Genetics Research Advisory Group, A first report to the NHS Central Research and Development Group on the new genetics, Department of Health, 1995

Service implications are discussed in the areas of: (1) The role of the regional genetics services (a role in training and education is stressed); (2) Maintenance of genetic registers; (3) The role of general practice in genetic services; (4) The appropriate organizational structure for a future era involving large-scale genetic testing and screening; and (5) Funding and patents.

The Genetics Research Advisory Group, The Genetics of Common Diseases. A second report to the NHS Central Research and Development Group on the new genetics, Department of Health, 1995

The report summarizes the current situation of clinical genetics services, discusses the financial implications of new genetics advances, and makes recommendations including: encouraging and coordinating research partnerships to carry out further research in genetic epidemiology, mutation detection techniques, full evaluation of genetic screening and its outcomes, and models of service organization; a systematic approach to the adoption of approved genetic screening schemes; the development of the role of primary care in genetic screening and counseling; education and training programs for professionals and the public; and a survey of existing genetic registers and their functions and effectiveness.

The Royal College of Physicians of London, Clinical Genetic services into the 21st century, Report of the Committee on Clinical Genetics, London, 1996 (www.rcplondon.ac.uk/pubs/index.html)

This report reviews the current situation and trends in: the nature of clinical genetic services; manpower in clinical genetics; the aims of medical genetics and the role of the clinical geneticist; the relationship between clinical genetics and other medical specialties. This report makes predictions and recommendations for the role of the clinical geneticist in the 21st century, the number of clinical geneticist and related posts that will be needed, the associated training requirements, and the organizational basis for clinical genetic services.

The Advisory Committee of Genetic Testing, Code of Practice and Guidance on Human Genetic Testing Services Supplied Direct to the Public, 1997 (http://www.open.gov.uk/doh/genetics.htm)

The Committee recognizes that medical practitioners in the National Health Service and private practice, and the commercial sector have roles to play in the provision of genetic testing services. The committee wishes to ensure that such services are delivered with the best interests of those tested in mind and that appropriate information and genetic consultation are available. Therefore, the Committee wishes to ensure that before introduction of services direct to the public, suppliers present their proposal to the Advisory Committee of Genetic Testing. The Committee will consider and monitor testing services in the light of the Code of Practice and Guidance.

The Advisory Committee on Genetic Testing, A report on Genetic Testing for Late Onset Disorders, 1998 (http://www.open.gov.uk/doh/genetics.htm)

The Committee sets out the issues to be considered before genetic testing for late onset disorders is offered and during the provision of such tests. Before any genetic test is used in clinical practice the scientific and clinical validity should be established. All laboratories providing genetic testing services should be closely linked with other genetic services, and be appropriately accredited for this.

Information on the disorder being tested for should be full, accurate and appropriately presented, in a clear and simple manner that is readily understandable. Appropriate support in preparation for and subsequent to genetic testing should be considered as part of the genetic testing process. In the case of presymptomatic genetic testing of healthy individuals, written consent should always be obtained. Tests for late onset disorders should not be supplied direct to the public.

The NHS, Commissioning in the new NHS commissioning services 1999-2000, London, 1998 (http://tap.ccta.gov.uk/doh/coin4.nsf/)

This document, issued by the NHS Executive, sets out the new arrangements for commissioning through Long Term Service Agreements. It includes arrangements for commissioning specialist services, which include clinical and laboratory genetic services.

The Royal College of Physicians, Commissioning clinical genetic services, Report from the Clinical Genetics Committee, London, 1998 (www.rcplondon.ac.uk/pubs/index.html)

This report sets out the requirements of a good clinical genetics service, and makes recommendations about how these requirements can best be met by commissioning bodies. It considers: the activities of clinical genetic services, the facilities required, the organization of services, commissioning mechanisms, management arrangements, costing, quality and performance indicators, and genetic services for common disorders.

The Royal College of Physicians, Clinical genetic Services. Outcome, effectiveness, quality, Report from the Clinical Genetics Committee, London, 1998 (www.rcplondon.ac.uk/pubs/index.html)

This report makes recommendations on the collection, storage, and retrieval of genetic data, the use of the outlined quality criteria in assessing effectiveness, and as the basis of service specifications, the need for multidisciplinary research to develop criteria for assessing the intangible outcomes and effectiveness of genetic counseling.

The Royal College of Physicians, Retention of Medical Records with Particular Reference to Medical Genetics, London, 1998

Genetic Interest Group, Confidentiality Guidelines, London, G.I.G., 1998 (http://www.gig.org.uk/docs/gig_confidentiality.pdf)

The purpose of these guidelines is to current practice in medical genetics in the UK with reference to individual confidentiality; to discuss ethical issues relating to the shared use of individual genetic information within families; to propose a framework to guide professionnals which formalises existing practice and to suggests a mechanism for resolving 'difficult' situations.

Genetic Interest Group, Guidelines for Genetic Services, London, G.I.G., 1998 (http://www.gig.org.uk)

The purpose of these guidelines is to help genetic and other service providers and commissioners, in partnership with service users, set and monitor standards, identify areas for improvement, devise strategies to develop and improve the services, and plan for the future. They cover: availability (service organization, staffing levels, funding), access and equity (referral arrangements, professional and public awareness, access for young people, people with disabilities, ethic minorities), partnerships with user and support groups and with other health professionals and services, good practice in providing information on genetic tests and diagnosis, good practice in genetic counseling (aims, content and scope, procedures, follow-up, confidentiality), long-term follow-up in families, standards for clinical and laboratory services monitoring and evaluation planning for the future.

The Clinical Genetics Society, The role of the clinical geneticist, 2000

This discussion paper produced by the Clinical Genetics Society documents the responsibilities of a clinical geneticist. Particular emphasis is placed on follow-up, support, coordination of health surveillance and services to extended families. Family involvement is the essence of the service which geneticists provide.

Laboratory Services for Genetics, Report of a working group to the NHS Executive and the Human Genetics Commission, 2000

The report recognizes the continuing role of laboratory genetics in service provision for single-gene disorders and recommends no immediate change to the current structure of the services, which are at present an integral part of the regional genetics centers and are often closely linked to university departments. However, it acknowledges that it is difficult to predict how laboratory services may need to evolve in the future if pharmacogenetic testing and testing for predisposition to common disease become a reality, and it recommends that the structure of the service should be kept under review for this reason. In assessing the effectiveness of current services, the working group found that the current regional basis for commissioning laboratory genetics services causes a number of problems, and recommends that the Department of Health should set up a national body to provide a 'strategic steer' on the commissioning of these services. In collaboration with the devolved administrations in the other countries of the UK, the Department of Health should consolidate an UK-wide genetic testing network to ensure the best provision for testing for very rare genetic diseases. The working group will re-convene in 2 years time to report on progress in implementing its recommendations.

United States of America Currently in the United States, genetic tests are regulated at the federal level through three mechanisms: (1) the Clinical Laboratory Improvement Amendments (CLIA); (2) the Federal Food, Drug, and Cosmetic Act; and (3) during investigation phases, regulations for the Protection of Human Subjects. Five organizations of the Department of Health and Human Services (DHHS) oversee genetic tests: the Centers for Disease Control and Prevention (CDC), the Food and Drug Administration (FDA), the Health Care Financing Administration (HCFA), the Office for Human Research Protections (OHRP), and the National Institutes of Health (NIH).

In addition to the Federal role, oversight of genetic tests is provided by states and private sector organizations. State health agencies, particularly state public health laboratories, have an oversight role in genetic testing, including the licensing of personnel and facilities that perform genetic tests. State public health laboratories and state-operated CLIA programs, which have been deemed equivalent to the Federal CLIA program, are responsible for quality assurance activities. States also administer newborn screening programs and provide other genetic services through maternal and child health programs.

The private sector provides oversight in partnership with HCFA and the CDC by serving as agents for the government in accreditation activities. The private sector also develops laboratory and clinical guidelines and standards. A number of professional organizations are involved in helping to ensure quality laboratory practices and in developing clinical practice guidelines to ensure the appropriate use of genetic tests. Professional organizations have also developed practice guidelines for specific disorders or groups of disorders (see http://www.faseb.org/genetics/).

American Society of Human Genetics, Statement on Clinical Genetics and Freedom of Choice, 1991 (http://www.faseb.org/genetics/ashg/policy/pol-07.htm)

The Society endorsed a proposal to modify restrictive abortion bills in order to protect the options of women at risk for bearing children with serious genetic or congenital disorders.

The Evaluation of Clinical Services Subcommittee, Great lakes Regional Genetic Group, Minimum Guidelines for the Delivery of Clinical Genetic Services, 1993

American Society of Human Genetics & The American College of Medical Genetics, Report: Points to consider: ethical, legal and psychosocial implications of genetic testing in children and adolescents, 1995 (http://www.faseb.org/genetics/acmg/pol-menu.htm)

This report focuses on genetic testing in response to a family history of genetic disease or to parents' request for genetic testing. This report is grounded in several social concepts: First, the primary goal of genetic testing should be to promote the well-being of the child. Second, the recognition that children are part of a network of family relationships supports an approach to potential conflicts that is not adversarial but, rather, emphasizes a deliberative process that seeks to promote the child's well-being within this context. Third, as children grow through successive stages of cognitive and moral development, parents and professionals should be attentive to the child's increasing interest and ability to participate in decisions about his or her own welfare. Counseling and communication with the child and family about genetic testing should include the following components: (1) assessment of the significance of the potential benefits and harms of the test, (2) determination of the decision-making capacity of the child, and (3) advocacy on behalf of the interests of the child.

US National Society of Genetic Counselors, Resolution on Prenatal and Childhood Testing for Adult-Onset Disorders, 1995 (http://www.geneclinics.org/profiles/webexcerpts/testing_resolution.html)

For adult-onset disorders for which the identification of gene carriers does not provide an avenue for therapeutic or preventive treatment in the prenatal or childhood periods, genetic testing must be carefully considered. In response to the unique nature of these disorders, the NSGC supports a series of recommendations: 'Clients considering a pregnancy or who have a fetus or child at-risk for an adult-onset genetic disorder should be made aware of clinically available testing technologies for that disorder. (…) Prenatal and childhood testing for adult-onset genetic conditions should always include genetic education and counseling. Genetic counseling for clients considering such testing should include exploration of the psychological/social risks and benefits of early genetic identification from both the parents and child's perspectives. When possible the child should be involved in the decision about whether or not to be tested. (…) Prenatal testing for adult-onset genetic conditions should be offered regardless of whether or not an affected fetus would be terminated'.

US National Society of Genetic Counselors, A position paper on Predisposition genetic testing for late-onset disorders in adults, 1997 (JAMA. 1997; 278: 1217-1220)

The Society recommends that professionals offering predisposition testing establish relationships with laboratories providing testing to optimize testing procedures and the clinical interpretation of test results. The Society does not take an explicit stance on commercial testing. The Society advocates responsible testing, whether commercial or noncommercial, for which persons receive appropriate education and counseling so that they can make autonomous informed decisions.

National Institutes of Health — Department of Energy group working on the ethical, legal and social implication of human genome research, Report: Promoting safe and effective genetic testing in the United States, 1997 (http://www.nhgri.nih.gov/ELSI/TFGT_final/)

The National Institutes of Health created a Task Force in order to review genetic testing in the United States and, when necessary, to make recommendations to ensure the development of safe and effective genetic tests. The report of the Task Force showed problems affecting safety and effectiveness of genetic testing in the US such as: validity and utility of predictive tests, laboratory quality, and appropriate use by health-care providers and consumers. On the basis of these findings, the Task Force made several recommendations to ensure safe and effective genetic testing. The Secretary of Health and Human Services followed up one recommendation by creating the Secretary's Advisory Committee on Genetic Testing (see below).

Council of Regional Networks for Genetic Services, Guidelines for Clinical Genetic Services for the Public's Health, 1997 (http://www.cc.emory.edu/PEDIATRICS/corn/news/pubs.htm)

These guidelines provide a framework to develop a state genetic services system. Concerning general facility and operational requirements, the guidelines state that 'the facility should be an identifiable unit in an accredited state or other medical school, a hospital, or a clinic accredited by the Joint Commission on Accreditation of Health Care Organizations. (…) Services should be available, accessible and culturally appropriate. (…) The center should develop and maintain an active program to monitor the quality of services provided. (…) Laboratories associated with the genetics unit should participate successfully in available proficiency testing programs. (…) No individual with a suspected genetic condition should be refused genetic services because of any disability or medical condition. State programs should provide support to those patients/families who are unable to pay'.

Statement. Professional disclosure of familial genetic information of the ASHG Social Issues Subcommittee on Familial Disclosure, 1998 (http://ns1.faseb.org/genetics/ashg/policy/pol-00.htm)

This report focuses on the potential conflict within the health-care professional–patient relationship when the patient refuses to warn at-risk relatives about relevant genetic information. Only exceptionally is a health care professional ethically permitted to breach confidentiality and as a legal matter ought to be privileged, that is, given a discretionary right to disclose genetic information to at-risk relatives without incurring liability provided certain conditions are met. Health care professionals should have an ethical duty to inform patients prior to testing as well as upon receipt of results that the information obtained may have familial implications.

American College of Medical Genetics, Standards and Guidelines for Clinical Genetics Laboratories, Second Edition, 1999 (http://www.faseb.org/genetics/acmg/stds/e.htm)

These voluntary standards are an educational resource to assist medical geneticists in providing accurate and reliable diagnostic genetic laboratory testing consistent with currently available technology and procedures in the areas of clinical cytogenetics, biochemical genetics, and molecular diagnostics. These standards establish minimal criteria for clinical genetics laboratories. The Standards should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. The accuracy and dependability of all procedures should be documented in each laboratory. This should include in-house validation and/or references to appropriate published literature. Specialized testing, not available to all laboratories, requires appropriate and sufficient documentation of effectiveness to justify its use. In determining the propriety of any specific procedure or test, the medical geneticist should apply his or her own professional judgment to the specific circumstances presented by the individual patient or specimen. Medical geneticists are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with these Standards. These Standards will be reviewed and updated periodically to assure their timeliness in this rapidly developing field.

American Society of Human Genetics and the American college of Medical Genetics, Joint Statement on Genetic Testing in Adoption, 2000 (http://www.faseb.org/genetics/ashg/policy/pol-36.htm)

The ASHG and the ACMG recommend the following. (1) 'All genetic testing of newborns and children in the adoption process should be consistent with the tests performed on all children of a similar age for the purposes of diagnosis or of identifying appropriate prevention strategies'. (2) 'Because the primary justification for genetic testing of any child is a timely medical benefit to the child, genetic testing of newborns and children in the adoption process should be limited to testing for conditions which manifest themselves during childhood or for which preventive measures or therapies may be undertaken during childhood'. (3) 'In the adoption process, it is not appropriate to test newborns and children for the purpose of detecting genetic variations of or predispositions to physical, mental, or behavioral traits within the normal range'.

Secretary's Advisory Commission on Genetic Testing, Enhancing the Oversight of Genetic Tests: Recommendations of the SACGT, 2000 (http://www4.od.nih.gov/oba/sacgt.htm)

SACGT has framed recommendations around the following five issues: (1) What criteria should be used to assess the benefits and risks of genetic tests? (2) How can the criteria for assessing the benefits and risks of genetic tests be used to differentiate categories of tests? What are the categories, and what kind of mechanism could be used to assign tests to the different categories? (3) What process should be used to collect, evaluate, and disseminate data on single tests or groups of tests in each category? (4) What are the options

for oversight of genetic tests and the advantages and disadvantages of each option? And (5) What is an appropriate level of oversight for each category of genetic tests?

Appendix B
Contributions

This document was reviewed by the ESHG Public and Professional Policy Committee (PPPC). Members of the PPPC were:

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The first draft was sent out to a wide range of people and organizations for consultation. It was reviewed critically by the following consultants:

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