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*R. Harris*[a]
*M. Reid*[b]

[a] Department of Medical Genetics University of Manchester, St. Mary's Hospital, Manchester, and
[b] Department of Public Health, University of Glasgow, UK

# Medical Genetic Services in 31 Countries[1]: An Overview

## Introduction

Our preliminary survey [1] of genetic services had difficulty documenting the availability of medical genetics services in the EU; indeed many countries had not yet formally recognised genetics as a medical speciality. Funding was therefore provided by the European Commission for a Concerted Action on Genetics Services in Europe (CAGSE) to set up a network of well informed clinical geneticists. With this network we have investigated the structure, workloads and quality of genetic services and the different responses made in each country to the new opportunities and challenges. Our aims were to promote international co-operation and to provide vital data to encourage medical genetics services consistent with the special needs of each country and the explicit wishes of consumers.

Medical genetics services are extending rapidly but even in well-resourced countries in northern Europe there is concern about rising demand, inadequate infrastructure and the ethical and social implications of the changes. For example when services for cancer genetics appear on the horizon, what are the limitations to offering such a service to a population? Where are the professional boundaries? What are the training needs of the providers? How do these problems compare with those of countries that have not had the opportunity to establish genetic services in the same way? Concern about cancer genetic developments may then be secondary to the problems of funding basic screening services, of obtaining state-of-the-art equipment and of building up a professional network of trained medical geneticists (see Stemerding et al., this supplement).

## Methods

To base the report on current practice (rather than future intentions), the members of the network (the 'authors') were asked to support their descriptions with detailed information placed in a standard framework of seven dimensions of health care [2]. We also used a SWOT (strengths, weaknesses, opportunities and threats) survey to obtain a general view of collaborators' concerns about the genetic services in their country.

*SWOT Survey*

There was consistency in these concerns and the most frequently cited items included the need for official recognition of the speciality of medical genetics, the need for adequate numbers of trained genetics staff and for a comprehensive national network of genetic centres. Factors cited as being important to access genetic services included comprehensive services regardless of patient income, problems of funding health services generally and regional inequalities of provision. Inadequacies were cited of teaching and training in medical genetics, quality assessment, trained counsellors, problems resulting from privatisation and inappropriate legislation. The results of the SWOT review emphasised the importance of genetic

[1] EU countries with Norway, Switzerland, some countries of the former Soviet bloc, Yugoslavia, Croatia, Slovenia, Israel, Cyprus and Turkey.

KARGER
E-Mail karger@karger.ch
Fax + 41 61 306 12 34



R. Harris
Department of Medical Genetics
University of Manchester, St Mary's Hospital
Hathersage Road
Manchester M113 0JH (UK)

centres to which reference was also frequently made in individual country chapters.

*Country Chapters*

Although the initial focus of CAGSE was on EU countries, it quickly became apparent that the comparative aspect would be strengthened by including those countries formerly in the Soviet bloc and others (Yugoslavia, Croatia, Slovenia, Israel, Cyprus, Norway, Switzerland and Turkey) which border the EU. The broader perspective throws into sharper relief the manner in which genetic services develop and the underlying prerequisites of such services. Although the report is comprehensive, including many countries, inevitably, given time and other constraints, a few countries have been left out, for example Estonia, Slovakia and Belorussia.

*Validation*

Ultimately, individual authors are responsible for the accuracy of the contents of their chapters, and we are very conscious of the challenges our authors have overcome to achieve consensus. Accuracy and balance were important goals and authors co-operated with great patience with the editorial committee who reviewed each chapter returning suggested revisions to authors, sometimes on several occasions. The chapters were also in 27 of 31 cases read and approved by senior 'validators' in each country, whose comments are summarised at the end of each chapter. This was to confirm accuracy and avoid too personal or too local a bias to the information. Validation letters frequently indicated the innovative nature of the processes used and the quality of the data, as the following examples illustrate.

The validator for Bulgaria, Professor P. Boyadjiev (now minister of health), stated:

'The demographic situation as well as organisation of medical care and genetic services are presented objectively. Obviously, a lot is to be done in the future in the field of genetic prevention both from educational and organisational point of view. Many ideas from the Chapter will be taken into consideration in the process of updating of the National Program for Genetic Prevention by the present and future Bulgarian Governments. Some financially dependent improvements are impossible at the moment, but the right ideas will be accomplished step by step in the future. The broader application of molecular methods for post- and prenatal diagnosis of genetic defects and genetic predisposition in both rare and common disorders is to be mentioned first amongst them.'

Professor G. Wolff, validating the German chapter, wrote:

'I fully agree with the content of this manuscript and approve its being published.'

The secretary of the Dutch Society of Human Genetics, Dr. Jacques Giltay, one of the validators for The Netherlands chapter, wrote:

'In my view it is a very accurate paper.'

The Polish validator, Dr. Anna Latos-Bielenska, stated:

'Human geneticists in Poland are most grateful to Prof. Zaremba for so accurate reflection of the real situation of medical genetics in our country. We are also grateful to [CAGSE] for this very important and useful idea of presentation of all genetic services in Europe.'

Marija Debevec, validating the Slovene chapter, wrote:

'On behalf of the Slovene Association of Human Genetics I acknowledge that the paper Genetic Services in Slovenia by Dr. Borut Peterlin presents the state of the art in human genetics in Slovenia.'

The Swiss validator, Professor D.F. Schorderet, in a letter to the author, Professor Pescia, stated:

'Thank you very much for the tremendous work you've done on surveying the Swiss Medical Genetics departments. I've read with great attention your manuscript and can validate your work. This, of course, represents a snapshot of our Medical Genetics institution as this field is continuously changing. Now that you have done all this work, I would like to invite you to present a short version of your draft to the next general assembly of the Swiss Society next fall. I'm sure they will have a great impact both in Europe and in Switzerland and I anticipate an important debate.'

Finally from Turkey, the comment from the validator was:

'I find it very beneficial and want to state that it is the only example from Turkey which has collated substantial data on this subject.'

*Note on Terminology*

Some CAGSE contributors have used the terms 'clinical geneticist' and 'medical geneticist' interchangeably but making a distinction where necessary by referring to genetically trained physicians and laboratory geneticists. Some prefer to use 'clinical geneticist' for both MDs and laboratory workers reserving 'medical geneticist' for medical graduates. The term 'genetic counsellor' is used to identify non-medical specialist nurses and psychosocial workers.

## Results

The task we set ourselves turned out to be challenging and rewarding. Each of the seven dimensions of genetic services actually warrants a study of its own and this report has its chief value in allowing initial comparisons to be made and broad principles identified.

### *Country Background*

Authors draw attention to the impact of major geographic, demographic, religious and political differences on provision of services. Geography plays a sometimes unrecognised role in fostering or hindering communications between professionals although computer linkages are rapidly bringing real improvement especially when geneticists use the Internet. Access to services by the populations may be constrained by geographical barriers, such as mountains or water, or simply by distance from major cities. The isolated valleys in Switzerland with genetic founder effects are an obvious example of the link between geography, genetics and populations. Norway and Greece both struggle with natural barriers that inhibit easy access to services. Countries vary too in their demographic structure. A comparison of vital statistics (table 1) emphasises the depressed state of some economies and in a few countries life expectancy has actually gone down in recent years. In the Ukraine for example, the comment was made (and echoed in several other countries) that the population was concentrating upon maintaining a basic level of existence and health issues were secondary to this more important concern. Life expectancy for men and women ranges from Russia where current rates according to EU data were 58 for men and 72 years for women and Turkey where the expectation is 65.7 and 70.3 years for men and women, respectively. These rates contrast sharply with 76.2 for men and 81.5 for women in Sweden. Infant mortality, the other major international comparative indicator, also varies very greatly from 44.4 deaths per 1,000 live births in Turkey to 3.9 in Finland.

### *Health Services Setting*

Across the 31 countries, one finds a diversity of forms of health service provision. Dual systems of state and private or insurance-supported systems are evident in the majority of countries. The availability of private laboratories and physicians working privately allows the population access to testing without going through the route of primary care referral. Privatised health care also forms a system of rationing the services and makes it harder to assess the services of a country since, almost without exception, few data are available about the volume provided within the private sector.

*From State Control to Market Principles*

The former Soviet bloc countries are working through the effect of the changes from state-controlled services to those run on market principles. This is reported to be liberalising in that it opens up additional routes to genetic services, private as well as some form of state or insurance based. However, the infrastructure is poor, and good access can have little impact if the genetic services cannot update their equipment and purchase new technology, and the staff do not have the facilities to update their knowledge.

*Primary Care*

'Primary care' for all medical problems is the first medical intervention and may be provided by specially trained general practitioners (GPs) or by a wide variety of specialists, including paediatricians, obstetricians and internists. Although little researched, many authors record the important role of the GP in some countries (notably the UK, The Netherlands and Denmark) as an important route and a 'gate keeper' to specialist care. However, the corollary is also noted, that a poorly informed GP can have a limiting effect on referrals for specialist care. Research is needed to assess the impact, consistency and knowledge base of primary care in the very different health care systems in Europe. (PrimGen is such a study and is part of CAGSE to be reported later.)

### *History and Medical Genetics*

The history of a country may continue to shape medical genetic services for many decades and in ways that are sometimes unavoidable. There is a powerful legacy of the Nazi regime and its gross eugenic excesses. German geneticists have sought to overcome this legacy but it is clear that it has had a lingering and profound effect on genetic services in Germany. For example, medical professionals are even more opposed than those in other countries to disease prevention as a primary aim of medical genetics. Furthermore, in Germany there is great concern to safeguard the confidentiality of genetic data and strong resistance to population genetic screening for carriers and to

**Table 1.** Statistical trends in Europe

| Country | Population mid 1995 | Area km² | Density persons/ km² | GNP per capita US$ | Life expectancy at birth | | Infant mortality/ 1,000 live births (1994–1995) |
|---|---|---|---|---|---|---|---|
| | | | | | male (1994–1995) | female (1994–1995) | |
| 1 Austria | 8,045,300 | 83,857 | 96 | 20,907 | 73.5 | 80.1 | 5.4 |
| 2 Belgium | 10,136,810 | 30,519 | 332 | 20,852 | 73.9 | 80.6 | 6.1 |
| 3 Bulgaria | 8,400,000 | 110,994 | 76 | 4,620 | 67.2 | 74.9 | 14.8 |
| 4 Croatia | 4,776,000 | 56,538 | 85 | 3,972 | 68.25[a] | 75.93[a] | 10.2 |
| 5 Cyprus | 732,700 | 9,251 | 79 | 11,652 | 75.3 | 79.8 | 8.5 |
| 6 Czech Republic | 10,330,759 | 78,864 | 131 | 21,502 | 72.5 | 77.8 | 5.3 |
| 7 Denmark | 5,228,000 | 43,094 | 120 | 26,000 | 72.4 | 77.8 | 5.4 |
| 8 Finland | 5,110,000 | 338,145 | 15 | 17,188 | 72.8 | 80.2 | 3.9 |
| 9 France | 58,142,852 | 551,500 | 105 | 19,955 | 73.8 | 81.9 | 4.9 |
| 10 Germany | 81,642,000 | 356,974 | 229 | 20,370 | 72.8 | 79.3 | 5.3 |
| 11 Greece | 10,458,711 | 131,990 | 79 | 11,650 | 75.0 | 79.9 | 7.7 |
| 12 Hungary | 10,229,000 | 93,030 | 110 | 6,607 | 64.8 | 74.2 | 10.7 |
| 13 Ireland | 3,582,000 | 70,285 | 51 | 16,431 | 73.2 | 78.7 | 6.3 |
| 14 Israel | 5,544,900 | 20,770 | 255 | 15,700 | 75.5 | 79.4 | 6.8 |
| 15 Italy | 57,301,000 | 301,268 | 190 | 19,536 | 74.8 | 80.9 | 6.4 |
| 16 Latvia | 2,515,600 | 64,589 | 39 | 3,261 | 60.2 | 73.1 | 18.5 |
| 17 Lithuania | 3,714,800 | 65,300 | 57 | 4,014 | 63.6 | 75.2 | 12.4 |
| 18 Netherlands | 15,459,006 | 41,526 | 372 | 19,341 | 74.6 | 80.4 | 5.5 |
| 19 Norway | 4,359,184 | 323,877 | 13 | 23,202 | 74.9 | 80.6 | 4.0 |
| 20 Poland | 38,588,000 | 312,683 | 123 | 5,478 | 67.6 | 76.4 | 13.6 |
| 21 Portugal | 9,920,000 | 92,389 | 107 | 12,841 | 71.5 | 78.6 | 7.4 |
| 22 Romania | 22,680,951 | 238,391 | 95 | 4,328 | 65.7 | 73.4 | 21.2 |
| 23 Russian Federation | 148,141,300 | 17,075,400 | 9 | 4,411 | 58.0 | 72.0 | 18.1 |
| 24 Serbia (Yugoslavia) | 10,544,278 | 102,173 | 103 | 440 (est) | 69.1[b] | 74.5[b] | 16.6 |
| 25 Slovenia | 1,987,505 | 20,256 | 98 | 10,725 | 70.3 | 77.8 | 5.5 |
| 26 Spain | 39,209,711 | 505,992 | 77 | 14,216 | 73.2 | 81.2 | 5.6 (est) |
| 27 Sweden | 8,827,000 | 449,964 | 22 | 18,201 | 76.2 | 81.5 | 4.2 |
| 28 Switzerland | 7,040,000 | 41,285 | 171 | 24,432 | 75.3 | 81.7 | 5.0 |
| 29 Turkey | 62,530,000 | 774,815 | 81 | 5,619 | 65.7 | 70.3 | 44.4 |
| 30 Ukraine | 51,531,500 | 603,700 | 85 | 2,383 | 62.8 | 73.2 | 14.7 |
| 31 United Kingdom | 58,260,000 | 244,100 | 239 | 18,360 | 74.2 | 79.5 | 6.2 |

EU data from *The Statistical Yearbook of the Economic Commission for Europe*, 1995 (http://www.unece.org/stats/trend/trend-h.htm). est = Estimated.

[a] 1989–1990

[b] 1992–1993.

There are some discrepancies with chapter data due to authors quoting different years.

genetic family registers. Based on the experience of the Nazi period, until recently, all aspects of medical genetics were opposed in Austria and the specialty is still not formally recognised.

The influence of Lysenko had a very damaging effect on genetics in the Soviet Union and associated countries in spite of the early origins (1932) of Russian medical genetics in the Institute of Medical Genetics in Moscow and the first medical genetics clinic in 1934. The Romanian author commented that it is difficult for foreigners to comprehend the unfortunate history of Romanian medical genetics when the 'iron curtain' separated Romanian physicians from their western colleagues and the only way to remain in contact with medical developments in the west was to emigrate. In contrast, the communist regime in the former Yugoslavia appears to have had a less inhibitory influence. For example, clinical genetics was encouraged by the first karyotype analysis performed in Croatia as early as 1959, the first genetic counselling clinics opening soon after. Amniocentesis was begun in 1969 and postgraduate courses in medical genetics were started in 1976. In Poland after the Second World War, the science of genetics was practically banned and medical students were indoctrinated with Soviet pseudoscience. However, authors report that such Soviet 'scientific authorities' as Lysenko and Lepieszynska were never taken seriously by most Polish scientists and after Stalin's death in 1953, the situation gradually improved.

*Politics*

The country chapters illustrate how politics has entered medical genetics. In addition to the effects of the Nazi and Lysenko eras, other influences have followed from the political commitment of the country and may have restricted the development of services by cutting off scientists and physicians from their counterparts in other countries. The east/west comparison is evident, where, for example, the Bulgarian author wrote that between 1944 and 1989 the country was cut off from modern (i.e. western) genetic theory and practice due to the communist regime. Authors from Croatia and Serbia wrote of the problems of running their services through recent wars. It is important not to forget that in western Europe, Portugal and Spain lived through dictatorships which, until fairly recently, made professional links with genetic services in other countries very difficult.

*Ethical Issues*

There are important ethical issues highlighted by the ways genetic services are provided in a wide range of countries. Basic ethical principles are embedded in tradition and institutions in every society. Identifying and using these principles for deliberations about problems in medicine and research has been a major conceptional resource in biomedical ethics. In her discussion of the seven dimensions of genetic services, Chadwick relates to the four-principles approach developed by Beauchamp and Children based upon autonomy, beneficence, non-maleficence and justice. In employing these principles for the CAGSE reports from the different participating European countries she derives the conclusion:

> 'The central question is the extent to which it is possible to mediate between autonomy and community, and in this specific context whether and how it is possible to pursue public health genetics without adopting policies associated with historical precedents now found unacceptable. Different countries will strike the balance between individual choice, public health and the good of the community in different ways.'

For the most part, genetic testing and services in the United States and western Europe have developed successfully over the past two decades by providing more options for avoiding inherited disorders. The interim report of the US Task Force on Genetic Testing of the NIH-DOE Working Group on Ethical, Legal and Social Implications of Human Genome Research states that this success is largely the result of genetic testing being undertaken in genetic centres or in consultation with geneticists and genetic counsellors. However, it is recognised that in the next few years the uses for genetic testing are likely to expand rapidly while the number of genetic specialists will remain essentially unchanged. This means that genetic testing decisions in the future will probably fall on providers who have little formal training or experience in genetics. The problems they will encounter in providing genetic tests are seldom encountered in other areas of medicine because genetic tests have the ability to predict risks of future disease although seldom does predictability approach certainty. The ultimate goals of human genome research and medical genetics are the treatment, cure and eventual prevention of genetic disorders. But treatment and cure lag behind the already available power to detect diseases or detect increased susceptibility to disease. Despite rapid advances in recent years, genetic testing is still in its infancy. The way from gene identification to clinical practice to public health application has still to be outlined and shaped. Therefore, emerging guidelines and recommendations on social and ethical issues in medical genetics and the provision of genetic services are important to ensure the development of safe and effective tests,

their delivery in laboratories of assured quality, and their appropriate use by health care providers and consumers not well educated in the inherent problems of genetic tests.

Ethical Boards exist primarily in northern European countries, for example Denmark and Sweden, and serve to highlight problems, but on occasion to inhibit procedures that are acceptable in other countries. The ending of funding for population cystic fibrosis carrier screening in Denmark is an example. In some other countries, ethical committees are at a less developed state. In 1994 in Latvia, for example, a Central Ethical Council at the Department of Health was established to deal with a broad array of ethical and consumer issues. Questions of medical/clinical genetics have not been discussed so far.

The chapter by Stemerding and his colleagues raises a new set of ethical issues to medical genetics through the introduction of cancer genetics. Although the majority of countries do not write about these new developments, they nevertheless represent an extension of genetic testing into the community. Even at this stage, however, the pressure to make the tests more widely available creates ethical and social problems (for the non-carriers as well as those diagnosed), in addition to creating dilemmas for those involved with the funding and management of the services.

## Seven Dimensions of Genetic Services in Europe

In the following pages, we summarise the authors' reports on genetic services arranged within the seven-dimensions structure [2], these are availability, access, life sustaining, non-harmful, effectiveness, state-of-the-art and consumer satisfaction. Here we have illustrated each dimension with a few examples but the individual chapters should be consulted for a fuller range of detail.

## Dimension 1: Availability

Under the heading 'availability', authors commented on the need for official recognition of the speciality, the numbers of professionals in medical genetics, facilities for genetic counselling, long-term care facilities, the provision of genetic laboratories and genetic screening. It is evident that there are major differences in availability within and among countries. The most commonly cited concerns, apart from lack of official recognition of the speciality, are inadequate numbers of trained genetics staff, lack of trained counsellors and the need for a comprehensive national network of genetic centres. In most countries, genetic services evolved in university departments and centralised genetic services generally remain in university/regional centres. The authors' descriptions demonstrate the remarkable extent to which medical geneticists in all countries agree the desired structure of genetic services. However, reference to table 2 and the following show that there are very great differences amongst CAGSE countries in what is actually available.

*Recognition*

CAGSE collaborators believe that official recognition of the speciality in each country and internationally (e.g. by the EU) is a crucial step towards improving the availability and access to the service. The reasons include the establishment and implementation of training programmes, the allocation of resources, recruitment to the speciality and relationships between medical geneticists and other specialities. It is therefore interesting to note that in only seven of the EU states is the speciality said to be recognised (Denmark, Finland, France, Germany, Netherlands, Sweden and UK). In Austria it is a subspeciality, but it is not recognised in Belgium, Greece, Ireland, Spain or Portugal. We do not have information on its official status in Luxembourg, or in Italy (where very large numbers, perhaps more than 400, of different subspecialities are said to be seeking recognition).

When comparing the stated recognition of medical genetics in individual countries there may be discrepancies between published sources. The procedures governing recognition of specialities in the EU are not easy to understand and depend on committees in the EU, in the individual states and on communication between them. These are currently beset by procedural problems that the EU 'Comitology Directive' is set up to address. In our work in connection with individual countries we have relied on authors' local knowledge and expect them to have confirmed this with their national health department.

*Roles*

The speciality of clinical genetics is rapidly changing because of new laboratory findings which require for their implementation trained cytogeneticists, molecular geneticists and biochemical geneticists who are committed to quality control and are engaged in developmental research. Genetics as a successful speciality depends upon interchange between laboratory geneticists, genetic physi-

cians and where possible, genetic psychosocial workers and genetic nurses. The roles of the genetic physicians include the diagnosis of inherited disorders and birth defects, elucidating recurrence risks, genetic counselling of family members and deploying clinical organisation, research and teaching expertise. Genetic physicians need a wide range of clinical skills relevant to disorders of all ages and all body systems. They need communication skills to be able to pass on complex ideas about risks and options and to help raise public awareness. Excellent relations are also essential with other medical specialities and primary care. In particular, new data on genetic risk factors for common diseases will require for their ethical and effective use many more professionals outside genetics with genetic literacy than now exist.

*Medical Genetics Physicians*

From data included in each chapter one can estimate that about 1,834 individuals in 28 countries were considered to be physicians working in genetic services. In a total population of 675 million there is a crude average of 2.7 per million population. However there are major differences in the distribution of medical geneticists (table 2). The number of 'physicians trained in genetics and working in genetic services' varies from 0.2 per million in Turkey to 2 or more in most of the rest and over 5 per million in Cyprus, Germany, Greece, Hungary and the Ukraine. However, the term 'genetic physician' remains poorly defined and precision will depend on acceptance of *minimum standards of training* to distinguish physicians with an interest who may be valuable colleagues but who lack formal training.

The data suggest that there are 355 physicians training in medical genetics in 24 countries. In these countries, the total number of 'trained' genetic physicians was 1,319, a ratio of 0.27 trainees to each physician. In some countries, e.g. Ireland, it is policy to rely on training in the UK or USA, until there is a critical mass of trainers within the country.

*Laboratory Scientists*

Problems with definitions also complicated measures of availability of genetics laboratory scientists. For example, when the term 'genetic counselling centres' was used it was not always clear whether clinical and laboratory diagnosis as well as genetic counselling were involved. In some countries, data suggested the involvement of medical graduates, PhDs and technicians in running genetics laboratories. In Germany and Spain, we were told that it was usual for an MD to both work in the genetic clinics and to perform diagnostic laboratory tests. In other countries, for example the UK, genetics laboratory staff are generally non-medical and laboratory reports are signed and issued independently, generally under the direction of PhDs. We were not always able to clarify the qualifications required by laboratory staff, and sometimes no distinction was made between trained cytogeneticists, molecular geneticists and biochemists and laboratory technicians or assistants. For these reasons, genetics 'laboratory staff' sometimes have to be considered as a whole and using this definition the data suggest that there are 2,375 professionals in 25 countries. The distribution ranges from 44 per million population in Cyprus to 1.2 in Turkey. Where it was possible to distinguish between cytogeneticists and molecular geneticists, the means are about equal at slightly more than 4 of each per million population (table 2).

*Genetic Counsellors (Genetically Trained Non-Medical Staff, Often Nurses, Clinical Genetic Co-Workers)*

The data suggest that there are about 800 non-medical genetic counsellors in 25 countries, with numbers ranging from 14.4 per million population in Hungary and 13.5 in Switzerland to none in France, Germany, Latvia, the Russian Federation, Slovenia, Spain and Turkey (table 2). In several countries, genetic counsellors provide a well-regarded and cost-effective extension to genetic services and in the UK an MSc course specifically for genetic counsellors has been established for 5 years and a similar course has recently been started in Greece. However, in other countries genetic counsellors appear to be regarded as a threat to doctors' professional perquisites.

*Genetics Centres*

Medical geneticists who work in multidisciplinary regional genetic centres with laboratory scientists, genetic counsellors and with academic colleagues comment on the advantages of close collaboration for both service and research. The UK, Netherlands and Belgium have clearly defined centres. France, which for historical reasons does not, has recently enacted legislation which will encourage the development of such centres. Genetic centres frequently make genetic services more widely accessible by setting up and supporting satellites in district or local hospitals.

**Table 2.** Availability and recognition of medical genetics: latest data provided by CAGSE chapter authors

| Country | Speciality officially recognised | Staff numbers | | | | |
|---|---|---|---|---|---|---|
| | | genetically trained physicians[a] | genetic physicians in training | cyto-geneticists | molecular geneticists | genetic counsel-lors[d] |
| 1 Austria | 1994[b] | 12 (1.5) | 3 (0.4) | 19 (2.4) | no data | |
| 2 Belgium | No | 34 (3.4) | 11 (1.1) | 22 (2.2) | 36 (3.6) | 22 (2.2) |
| 3 Bulgaria | 1985 | ca. 30 (3) | | 36 (4.2) | | 28 (3.3) |
| 4 Croatia | No | 5 (1)[e] | 1 (0.2) | 14 (2.9) | 5 (1.1) | 8 (1.7) |
| 5 Cyprus | No data | 4 (5.5) | 2 (2.7) | 10 (13.7) | 22 (30.1) | 6 (8.2) |
| 6 Czech Republic | 1980 | 46 (4.5) | 6 (0.6) | 46 (4.5) | 10 (10) | 33 (3.2) |
| 7 Denmark | 1996 | 20 (3.9) | 5 (1) | | 130 (25) | |
| 8 Finland | 1981 | 17 (3.3) | 10 (2) | 9 (1.8) 6 in training | | 23 (4.5) |
| 9 France | 1995 | 125 (2.2) | 20 (0.35) | 200 (3.5) | 100 (1.8) | 0 |
| 10 Germany | 1992 | 441 (5.4) | | no data | | 0 |
| 11 Greece | no | ca. 60 (5.7) | | no data | | 2 (0.2) |
| 12 Hungary | 1978 | 56 (5.5) | 20 (2) | 19 (1.9) | 20 (2) | 147 (14.4) |
| 13 Ireland | no | 2 (0.6) | 0 | 13.6 (3.8) | 8 (2.2) | 1 (0.28) |
| 14 Israel | 1986 | 17 (3.2) | 3 (0.6) | 32 (6) | 19 (3.5) | 16 (3) |
| 15 Italy | 1940 | | | no data | | |
| 16 Latvia | –[c] | 7 (2.7) | 0 | 7 (2.7) | 4 (1.5) | 0 |
| 17 Lithuania | 1991 | 8 (2.2) | 2 (0.54) | 2 (0.54) | 7 (1.9) | 4 (1.1) |
| 18 Netherlands | 1987 | 55 (3.5) | 20 (1.2) | 25 (1.6) | 25 (1.6) | 6 (0.4) |
| 19 Norway | 1971 | 11 (2.6) | 7 (1.6) | | no data | 3 (0.6) |
| 20 Poland | 1998 | 96 (2.4) | 4 (0.02) | | 139 (3.6) | 8 (0.04) |
| 21 Portugal | no | | | no data | | |
| 22 Romania | 1997 | | | no data | | |
| 23 Russian Federation | 1988 | 300 (2.02) | 60 (0.4) | 215 (1.5) | 50 (0.3) | 0 |
| 24 Serbia | no | 11 (1) | 11(1) | 25 (2.4) | 10 (1) | 6 (0.6) |
| 25 Slovenia | no | 6 (3) | 2 (1) | 7 (3.5) | 15 (7.5) | 0 |
| 26 Spain | no | 75 (1.9) | ? | 84 graduates, 145 technicians | | 0 |
| 27 Sweden | 1991 | 16 (1.8) | 4 (0.5) | 10 PhD, 80 technicians | | 1 (0.1) |
| 28 Switzerland | no | 15 (2.2) | 10 (1.5) | 17 (2.5) | 13 (1.9) | 93 (13.5) |
| 29 Turkey | 1990 | 14 (0.2) | 25 (0.4) | 49 (0.8) | 28 (0.5) | 0 |
| 30 Ukraine | 1993 | 272 (5.1) | 74 (1.4) | | 133 (2.5) | 286 (5.4) |
| 31 United Kingdom | ca. 1970 | 79 (1.4) | 55 (0.95) | 408 (7) | 101 (1.7) | 107 (1.8) |

[a] Values in parentheses – per million population.
[b] Reported as 'subspeciality'
[c] Reported as 'additional (non-basic) medical speciality'.
[d] Non-medical specialist nurses, psychosocial workers.
[e] Reflects those working only in diagnostic services. Number working in research is significantly higher.

## Dimension 2: Access

*Factors Influencing Access*

Many country authors noted that *in theory* there were no barriers to access to the services and that the respective health system of their country allowed 'free access'. However, the statement was often qualified with a description of a number of factors which played a part in limiting or encouraging access to the genetic services. Under this heading authors considered the influence on access to genetic services of primary care referrals, religious, ethnic, legal or other barriers to abortion, geographical features limiting access, the cost of genetic services, whether there is full reimbursement for genetic services and the extent to which private genetics laboratories have been established. Thus a discussion of access indirectly involves aspects of other dimensions discussed within these chapters including service funding, professional training (and knowledge) and links with consumers.

Table 3 summarises the main factors which the authors have reported *limit* access to services. Funding issues feature both directly in western and some of the formerly eastern European countries (Spain, Portugal as well as Croatia, Russia, Latvia), with lack of funds contributing to a situation of too few specialists and the lack of new posts in genetics. Some authors have also noted the problems caused by administrators who do not understand the services, while in a few instances authors have noted that funding limits the purchasing of basics, new reagents and tests (e.g. Ukraine). These factors too have an impact upon the uneven 'spread' of services and centres, while geography can also play a role in fostering or hindering communications between professionals. Access to services by the population may be constrained by geographical barriers, such as mountains or water, or simply by distance from major cities. Unequal access is noted in many countries with a tendency for genetic centres to be located in large cities or at least unevenly distributed throughout the country. Lack of knowledge and understanding of genetics by 'non-geneticist' doctors is cited in many country chapters as an important reason for poor access of patients (for example in Germany, Latvia, Russia, and Slovenia), with the implicit recognition that service use depends on satisfactory professional referrals. Some authors have underlined the importance of good professional links, reporting that poor professional networking inhibits good access. However, patients' lack of knowledge of the services is also a significant factor noted by the majority of contributors. This is not a straightforward issue; health insurance may limit the tests that patients may undergo free of charge, while if the patient is not covered they may not be able to pay for certain tests. Although this is seldom researched, authors have noted explicitly that some patients gain greater access to services because of their better education or knowledge. In some countries, patients living in rural areas may be doubly disadvantaged, by their own lack of awareness of the services available in the cities and by rural professionals' lack of knowledge.

Authors have also mentioned the role that other factors play in affecting access; included here are religion (Austria, Ireland, Portugal), the high prevalence of certain genetic conditions (Greece) and the particular problems of access raised by ethnic minority groups who may have difficulties of language, comprehension of the health services or a lower likelihood of health insurance (Germany, Netherlands).

Belgium and Finland offer interesting examples of access. Belgian genetic services have interesting if paradoxical features. The cost of testing is low and almost fully refundable, so that access is, in principle, freely available to every individual and the whole population should be able to benefit. Even though medical, human and molecular genetics are not recognised specialities in Belgium, genetic services are officially restricted to eight licensed centres and no other institution or laboratory will be refunded for genetic activities. As there is no primary care referral system, it is not clear how patients needing genetic services are identified and referred to genetic centres.

In Finland there is a nationwide system of health centres which, with their associated maternity and child care units, look after virtually the whole population. A tiered system allows for the identification of genetic problems and for their reaching an appropriate specialist by rising through the system to general hospitals and then to medical genetic units. Because patients can also contact genetic services directly (without being referred), the chances of everyone having access to the services are said to be very good.

No country appears to be able to offer complete access to the full range of possible genetic services and introducing, encouraging or transferring to private sector services is one way of limiting or 'rationing' the demand, but not of obtaining equity of access.

*Prenatal Screening and Diagnosis*

This area was not a specific focus of CAGSE but has been written about in 'The diffusion of four prenatal screening tests across Europe' [3] and more recently in

**Table 3.** Factors reported to affect access to medical services in CAGSE countries

| Country | Poor funding | Administrator's poor knowledge | Poor professional networks | Centres mainly in cities | Geographically difficult | Doctors lack knowledge | Patients lack knowledge | Other factors |
|---|---|---|---|---|---|---|---|---|
| Austria | | × | | | × | × | | religion |
| Belgium | | | | | | | | insurance – number of tests |
| Bulgaria | | | | | | × | | |
| Croatia | × | | | | × | × | | |
| Cyprus | | | | × | | | | politics |
| Czech Republic | | | | | | × | × | |
| Denmark | × | × | × | | | | × | |
| Finland | | | | × | | | × | |
| France | | | | | | | × | |
| Germany | | | | × | | × | × | immigrants – rare diseases |
| Greece | | | | × | | | | high frequency of specific diseases |
| Hungary | × | | | × | | × | × | |
| Ireland | | | | | | | | religion |
| Israel | | | | | | | × | patients have to pay for some tests |
| Italy | × | × | | | | × | × | not homogeneous distribution of services |
| Latvia | × | × | × | | | × | × | |
| Lithuania | × | | | | | × | | patients have to pay for some tests |
| Netherlands | | | | | | | × | ethnic minority women may use services less; religion |
| Norway | × | | × | × | × | × | × | legal restrictions |
| Poland | × | × | × | × | | × | | religion. Patients have to pay for some tests |
| Portugal | × | | | × | | × | × | |
| Romania | | | | | | | | |
| Russia | × | | × | × | | × | × | |
| Serbia | × | × | × | | | | × | |
| Slovenia | | | | | | × | × | |
| Spain | × | | | × | | × | × | |
| Sweden | | × | × | | × | × | | |
| Switzerland | | | | | | | × | language |
| Turkey | | | | × | | × | × | |
| Ukraine | × | | | | | | × | |
| United Kingdom | × | | | | | × | | ethical issues cause hesitancy |

'Prenatal diagnosis in Europe' in the proceedings of the EUCROMIC workshop [4]. However, in the present study some authors have drawn attention to severe limitations in access to prenatal diagnosis. For example, in Bulgaria an underreferral rate is well documented and only 600 pregnancies in women aged over 35 years were tested for chromosome anomalies in the last 12 years although the *annual* rate of pregnant women in this age group exceeded 2,000. By contrast, the Czech Republic has widespread public awareness and a favourable attitude towards services.

*Neonatal Screening*

Neonatal screening for phenylketonuria (PKU) and congenital hypothyroidism (CH) are nearly universal in Europe and only Romania has no such programmes at present (table 4). In a few countries, genetic centres are actively involved in running screening but in most their role is limited.

*Screening for Carriers and for Late-Onset Genetic Disorders*

Table 5 illustrates several important themes. Presymptomatic and predictive genetic testing for late-onset genetic disorders occurs mainly in the affluent north-west of Europe. There is professional reluctance to be associated with population screening in a few countries, including Germany (a clear example) but also The Netherlands. In contrast, there are intensive carrier screening programmes allied to prenatal diagnosis for example for thalassaemia in countries with Mediterranean populations.

*Therapeutic Abortion*

Access to therapeutic abortion is variable as a means of avoiding genetically abnormal live births, although in most countries, termination is permissible until 12 weeks and, when an abnormality is demonstrated, until 22 weeks or later (e.g. UK, until full term). Access to prenatal diagnosis in Europe has been documented in the proceedings of a recent EUCROMIC workshop [4].

*Less Common Disorders*

In some countries, diagnostic tests may not be available for less common disorders. Access to these tests only becomes possible when patients or samples are sent to other countries that have better services for the tests. The de facto international network operates reasonably well when funds and hard currency are available to pay for the tests, but in some countries there may not be sufficient resources to pay even for the cost of transportation.

*Ethnic and Cultural Influences*

Countries vary greatly in their ethnic mix, a feature that becomes particularly significant when considering inherited conditions that are found more commonly in certain populations. These include thalassaemia in Greece, Cyprus and Italy, and Tay-Sachs and Canavan disease in Israel. The extreme heterogeneity of the populations of the Russian Federation is associated with enclaves of rare genetic disorders. Such variables and special needs throw an additional load on genetic services which have often set up successful comprehensive services as typified by the Sardinian, Cypriot and other thalassaemia programmes. In Turkey, one-third of the population is under the age of 15, and 22% of marriages are consanguineous, with 66.3% of those being first cousin marriages (this proportion has decreased by 10% in the last 30 years due to public health initiatives). The Israeli author also notes that a high proportion of Bedouin marriages in Israel are consanguineous.

*Genetic Risk Factors for Common Diseases*

A major life sustaining influence, which will be exerted by genetics, is only now beginning with the discovery of genetic risk factors for common diseases and their introduction into health care. Stemerding, Koch and Bourret in this volume describe the emergence of cancer genetic services in European health care, which herald 'a complex series of challenges' to the health care system. The establishment of multidisciplinary cancer family clinics offering specialist counselling, and the establishment of registries of individuals at risk, are generally perceived as necessary steps in this direction. Future models of cancer genetic services also emphasise the role of GPs in identifying women with a family history of breast cancer and in educating and counselling them about their personal risk factors.

*Religion*

Religion is a curiously unpredictable factor. Its influence is seen in Ireland, where strict adherence to Roman Catholicism inhibits the development of those medical genetic services that have at their root abortion for avoiding severe birth defects. Recent developments in Ireland have liberalised the situation to the extent that prenatal diagnosis is now increasingly accessible, but women must travel to other countries, mainly the UK, for legal abortion of abnormal fetuses. Elsewhere, access to services may be limited, not by law, but by failure to refer by doctors who have strong religious beliefs. Doctors may explicitly opt to be 'conscientious objectors' and presum-

**Table 4.** Newborn screening: data provided by CAGSE chapter authors

| Country | Newborn screening |
|---|---|
| Austria | PKU, CH, galactosaemia, local screening for CF outside approved programmes |
| Belgium | PKU, CH, galactosaemia and other treatable diseases |
| Bulgaria | PKU and galactosaemia in 80% of all newborn; CH is carried out in a separate lab |
| Croatia | PKU from 1978, CH from 1984 |
| Cyprus | 95% neonates tested for PKU, CH and DMD in newborn boys |
| Czech Republic | all neonates tested for PKU and CH |
| Denmark | national PKU and CH; 21-hydroxylase deficiency pilot study |
| Finland | CH with almost 100% uptake |
| France | PKU, congenital adrenal hyperplasia and CH; national programme for cystic fibrosis at birth ran until 1992 and still active in some regions; sickle cell anaemia in children according to ethnic origin |
| Germany | PKU, galactosaemia, CH in all states. Biotinidase deficiency in approximately 1/3 newborns screened. Pilot study for 21-hydroxylase |
| Greece | PKU, CH, G6PD deficiency and galactosaemia covering more than 95% of newborns in Greece |
| Hungary | PKU, galactosaemia, CH and biotinidase deficiency; congenital dislocation of the hip which was the commonest congenital abnormality |
| Ireland | PKU, CH and galactosaemia centrally |
| Israel | PKU, CH and congenital malformations on every newborn |
| Italy | thalassaemia, PKU, galactosaemia, congenital CH, according to the different regional laws |
| Latvia | PKU whole of Latvia from 1987, CH from 1995 |
| Lithuania | PKU and CH began in 1975 and 1993, respectively; CH discontinued for 8 months in 1996 due to funding |
| Netherlands | PKU and CH with a very high coverage |
| Norway | PKU 1967, nationally from 1978; CH started in 1979: parents' right to newborn screening if early diagnosis improves the prognosis |
| Poland | PKU and CH span the whole country |
| Portugal | PKU and CH covers over 95% (1994) of the newborn population |
| Romania | currently no prenatal or postnatal screening programmes |
| Russia | PKU and CH routine in European Russia, but vary in other parts of the country; 74.7% PKU, 54.3% CH |
| Serbia-Montenegro | screening programmes for PKU and CH started 1976 ceased because of war and lack of money |
| Slovenia | PKU and CH for whole population; cystic fibrosis available 1995–1996 through research projects |
| Spain | PKU and CH cover practically 100% of the population |
| Sweden | PKU, galactosaemia, CH and the adrenogenital syndrome with acceptance close to 100% |
| Switzerland | PKU, CH and galactosaemia nationwide |
| Turkey | PKU nationwide started in 1983; CH added in 1991 |
| Ukraine | PKU nationwide |
| United Kingdom | PKU nationwide |

DMD = Duchenne muscular dystrophy; G6PD = glucose-6-phosphate dehydrogenase; CH = congenital hypothyroidism

**Table 5.** Screening for carriers of recessives and for late-onset genetic disorders: comments made by CAGSE chapter authors

| Country | References to screening (other than prenatal and neonatal) |
|---|---|
| Austria | No countrywide population screening programmes. |
| Belgium | Presymptomatic counselling and testing of individuals for HD, spinocerebellar ataxia, familial amyotrophic lateral sclerosis and families with increased risk for other late-onset diseases such as malignancy, psychiatric diseases or cardiovascular diseases; implemented for colon and breast cancer. |
| Bulgaria | No data available. |
| Croatia | No population screening for carriers of recessive diseases is provided. |
| Cyprus | An efficient nationwide school screening programme for thalassaemia allows for premarital carrier diagnosis. |
| Czech Republic | Predictive tests for hereditary disorders are not done in the Czech Republic, except to close relatives in families with CF and HD. |
| Denmark | CF carrier screening as a pilot project 1990–1992. |
| Finland | Only research in families with hereditary non-polyposis colorectal cancer, breast/ovarian, multiple endocrine neoplasm. Carrier screening is not routinely done. Pilot programmes for aspartylglucosaminuria, infantile neuronal ceroid lipofuscinosis and fragile X. |
| France | Population screening is available only in neonatal period but guidelines on predictive testing published. |
| Germany | Population screening for carrier status in adults 'not actively promoted'; German Society for Human Genetics opposes testing of minors. |
| Greece | Population screening for carriers in some places; haemoglobinopathy screening/counselling free of charge. |
| Hungary | High rate (about 52%) of death due to cardiovascular diseases led to general and specific cholesterol screening and MED-PED programme to detect familial hypercholesterolaemia. |
| Ireland | Apart from neonatal, no population screening. |
| Israel | For all Ashkenazi and Moroccan Jews at reproductive age, screening for Tay-Sachs and Canavan disease carriers, and β-thalassaemia in Jews from Kurdistan and Arabs. Cystic fibrosis carrier screening since 1994. Ultra-Orthodox Jewish Ashkenazi marriages 'arranged' to avoid Tay-Sachs disease, CF and Canavan disease. |
| Italy | Population screening regulated by regional laws. Sardinia: thalassaemia screening. |
| Latvia | Only close relatives in CF families. |
| Lithuania | Population screening for carriers of recessive disorders is not performed. |
| Netherlands | No programmes for carrier screening for CF or haemoglobinopathies. |
| Norway | Diagnostic genetic testing permitted without restrictions if older than 16 years but if presymptomatic, predictive or for carrier detection, law states that there must be counselling before, during and after test. |
| Poland | No carrier screening for CF. |
| Portugal | Screening for CF carriers is offered only to families with affected members but presymptomatic tests may be given without genetic support; haemoglobinopathies screening according to WHO guidelines. |
| Romania | No postnatal screening programmes or population screening for carriers of recessive disorders. |
| Russia | No data available. |
| Serbia-Montenegro | No data available. |
| Slovenia | No population screening and predictive tests not routinely in use but research on genetic factors in atherosclerosis, risk factors for coronary heart disease and cerebral vascular insult as well as genetic analysis of breast cancer is currently underway. |
| Spain | Presymptomatic testing and carrier detection in some genetic units/centres for individuals at risk of recessive and late onset disorders. Experience is very limited of predictive testing for cancer but research-based testing for breast and colon. |
| Sweden | Predictive testing for neurogenetic and genetic cancer disorders. |
| Switzerland | Predictive testing for cancer susceptibility genes is being developed. |
| Turkey | Screening for haemoglobinopathies in primary and high school with premarital screening where high incidence of HbS; centre now established for cancer genetics. |
| Ukraine | After Chernobyl, obligatory annual preventive examination to identify high-risk groups and early cancers. Inherited tumour syndromes are registered to identify high-genetic-risk groups. |
| United Kingdom | Predictive testing for HD and hereditary cancers widespread but not on population base; CF carrier testing on research base. |

CF = Cystic fibrosis; HD = Huntington disease.

ably refer patients to colleagues. However, some countries with a strong religious foundation, for example Italy, have embraced medical genetic services fully.

## Dimension 3: Life Sustaining

Basic economic and demographic characteristics including adult life expectancy and infant mortality are important measures of the state of public health, and form an essential background to the study of genetic services (table 1). With historical and other factors they must be

taken into account if one is to avoid making inappropriate comparisons between the north west of Europe, the south of Europe and the countries of the ex-Soviet bloc. Genetic services are particularly undervalued in countries struggling for economic reform.

## Dimension 4: Non-Harmful

*Quality Control*

The extent to which countries have worked out systems of quality control is an important indicator of their service development. Although the majority of authors report that measures of *external* quality control do not exist, in countries where central support has been positive, external genetic laboratory quality control has begun, for example in the UK, Ireland, Netherlands and Denmark. There is at least one national study of genetic counselling by non-geneticists in the form of UK national confidential enquiries [5] which involve the ascertainment of all cases of marker disorders within a defined period and are modelled on British national confidential enquiries into maternal and perioperative deaths. This methodology may be slower to reach CAGSE countries in which obstetricians, surgeons and anaesthetists have not yet set up comparable confidential enquiries into maternal and perioperative deaths.

*Legislation*

The extent of legislative involvement in services is manifest in many ways. In a number of western European countries where genetics is well established, a legal framework exists within which the services operate. Norway is perhaps an extreme case where the law requires counselling for presymptomatic, predictive or carrier testing before, during and after the test. A further Act prohibits the results of testing being made available to anyone other than the individual concerned. Thus employers and insurance companies are prevented from obtaining results for their own purposes. However, in other countries there is no legal framework concerning such tests and the delivery of services; test results may be issues without counselling and may be accessible to third parties. In Bulgaria, for example, there are no constraints on prenatal sex selection. In Russia there is little legislation regulating genetic testing and gene therapy; currently laws are being drafted concerning in vitro fertilisation, gene therapy and research involving eggs and embryos. In UK the Human Fertilisation and Embryology Authority has statutory powers, while other influential advisory bodies exist including the Advisory Committee on Genetic Testing.

## Dimension 5: Effectiveness

*Outcomes*

Medical geneticists do not use outcome measures based on prevention of handicapping disorders by prenatal diagnosis and selective abortion and believe that 'non-directive counselling and the provision of choices' are essential but have benefits that are too difficult to assess routinely. This area has been comprehensively reviewed in a Royal College of Physicians of London report [6] that recommends using proxy measures of quality including scope, accessibility, responsibility, quality of clinical care and laboratory services. Although these proxies are implicit within the seven dimensions of health care, many country authors state that quality is difficult and sometimes impossible to comment on at present. Quality can only be judged when geneticists have access to reliable information about how services are delivered in terms of clinical care, whether test results are accompanied by counselling the long-term outcomes and so on. Few countries have access to this kind of data. The overall view is that the quality of genetic services differs considerably among countries and also within them. The increasing role of private health care makes assessment of quality, no matter how measured, even more difficult. In some countries, the major concern is to provide any service at all, and issues of quality become secondary.

*Uptake*

The country chapters document the whole spectrum of uptake of services from the few countries where routine amniocentesis of older mothers is a luxury to those few countries at the other end of the spectrum where prenatal screening and diagnosis are widely available and are usually taken up. In some countries, predictive screening for a range of cancers is already in place and exposed to overwhelming demand from families. Uptake is influenced by capacity, not only of laboratories but also of the overall services including staffing, and this capacity in turn is a result of the availability of funds. Genetics is a rapidly expanding scientific discipline and each new development has implications for the level of technology required and the expertise of the staff. In many countries, research funding will only permit development, but not routine service use. Examples include the development of chorionic villus sampling and, more recently, fluorescent in situ hybridisation analysis.

Uptake is influenced by other factors including the 'culture' of a country and the way services are resourced. For example, the author from the Ukraine notes that the

**Table 6.** Genetic family registers: comments made by CAGSE chapter authors

| Country | Comments |
|---|---|
| Austria | There are no genetic registers in Austria. |
| Belgium | National register of about 200 FAP families: some data collected for EUROCAT congenital abnormalities register. |
| Bulgaria | Registration of congenital malformations in newborn. |
| Croatia | Apart from the Croatian Cancer Register there are no national registers. |
| Cyprus | No data available [assumed registers must exist for thalassaemia families, ed.] |
| Czech Republic | Genetic departments have their own registers of patients and their families for genetic counselling. State register of congenital malformations. |
| Denmark | All FAP families, as well as some other inherited tumour syndromes including Von Hippel-Lindau, neurofibromatosis, retinoblastoma. |
| Finland | Confidentiality laws preclude official genetic family registers although scientists may have their own registers on families included in their studies and cancer register run by Cancer Society of Finland. |
| France | Two genetic registers in France: CF and sickle cell anaemia; also four population-based registers of birth defects including chromosomal anomalies and genetic syndromes recognisable at birth and surveying 25% of the births in France. |
| Germany | There are no official genetic family registers. |
| Greece | There are no general genetic, hereditary cancer, cytogenetic, abortion or congenital anomaly registers. |
| Hungary | Congenital abnormality registry; National Registry of Childhood Cancer. |
| Ireland | No data available. |
| Israel | There is a national register for congenital malformations and for Down syndrome. Other registers for genetic disorders are planned. |
| Italy | The National Institute of Health Care yearly statistics on the most important congenital malformations, and genetic family registers kept by laboratories of patients referred. |
| Latvia | Latvian State Register of birth defects which reports regularly on registered congenital malformations. All cases of diagnosed genetic disorders are also registered. |
| Lithuania | Lithuanian Registry of Inherited Diseases and Congenital Anomalies (LIRECA). |
| Netherlands | Registration of congenital anomalies through national obstetrics and neonatal registries, and EUROCAT. Several national registries of individual disorders, such as CF and retinoblastoma and long-term follow up of patients with a high risk of developing tumours is performed by a special foundation. |
| Norway | A register for autosomal dominant polyposis families exists within the Cancer Registry, and routines are being developed for information to all at-risk persons about clinical follow-up and genetic counselling. |
| Poland | Polish register of congenital malformations established for western and central Poland 1997. Oncological register of hereditary tumours in Szczecin. |
| Portugal | There are no genetic registers. |
| Romania | National registers for a few diseases e.g. sickle cell disease, β-thalassaemia and congenital malformations. Regional registers for hereditary colorectal cancer. |
| Russia | All families at high risk of inherited disorders are registered at all levels of the medical genetics service and placed on the relevant local computerised registers of the Regional Medical Genetic Centre. |
| Serbia-Montenegro | The national congenital malformation registry is in Novi Sad genetic centre. From 1987 register of CF families. |
| Slovenia | Significant developments in terms of improved genetic counselling or prevention are communicated to patients with an increased risk of severe genetic disorders via the register kept at the Division of Medical Genetics. A cancer family clinic is being developed and the Cancer Registry of Slovenia has maintained a database of cancer patients since 1950. |
| Spain | One national and 4 regional registers of congenital malformations. Departments have own registers. |
| Sweden | 7 compulsory registries: Medical Birth Registry, Congenital Malformations, Central Unbalanced Cytogenetic Registry Cancer Registry. Cause of Death Registry, Paediatric Cardiac Malformation Registry, Prenatal Malformation Registry. Also numerous national and regional registries administrated by groups of doctors for research and clinical follow-up such as a Huntington Disease Registry and Colon Polyposis Registry. |
| Switzerland | Some cantons have registers for familial cancers particularly FAP and retinoblastoma. |
| Turkey | No genetic family registers reported. |
| Ukraine | District counselling institutions also register families with hereditary pathologies and congenital defects. Inherited tumour syndromes are registered at the Interregional Medical Genetic Centre and the data are used to identify high-genetic-risk groups. |
| United Kingdom | The first genetic registers established in the 1970s with increasing work involving individuals requiring regular review; registers regarded as essential tools for genetic services. |

FAP = Familial adenomatous polyposis; CF = cystic fibrosis.

government makes prenatal testing seem less necessary by relieving families of the burden of handicapped children by making institutional provision. In Bulgaria, the author comments that only 2,000 of the approximately 30,000 children living in special homes have no families. A different 'uptake' issue is illustrated in Latvia where it is reported that maternal serum AFP screening for neural tube defects and Down syndrome was available to all pregnant women in 1991–1992 but is now restricted to those who can pay (about 2,000 analyses per year). Prenatal diagnosis is still relatively uncommon because of poor availability of laboratory reagents and equipment as well as poor public awareness. Authors from many countries reported problems with re-equipment and procurement of reagents and kits.

In contrast, prenatal screening is reported to be used increasingly where there is a predominant genetic disorder like thalassaemia, or where consanguinity is common (e.g. Turkey). Several authors comment that the availability of prenatal screening may encourage older mothers to become pregnant.

*Genetic Registers*

Table 6, which summarises data provided by authors, includes formal or informal registers in individual departments as well as national 'registries' of congenital malformations, and illustrates the variable emphasis given to registers in service provision. Genetic family registers involve records on *kindreds* and are disease specific (e.g. Duchenne, Huntington disease, multiple polyposis coli) and differ from many disease or procedure registers, e.g. registers of congenital malformations or prenatal diagnosis. Genetic family registers have long been used in the UK [7] and are considered a logical approach to long-term responsibility for counselling families with severe autosomal dominant or X-linked disorders, particularly when young people approach reproductive age and new genetic tests are developed. There is believed to be a particular advantage to the families who can consider the tests and choose their reproductive option before the pressure of pregnancy arrives. The potential value of genetic family registers could greatly increase as genetic risk factors for common diseases become clinically useful and require tracking through families. The chapter by Stemerding, Koch and Bourret illustrates the use of genetic family registers to aid the management of hereditary cancers.

Although most medical genetics departments have details of families in their records, few countries have adopted genetic family registers as a routine service tool. In Germany, formal genetic family registers do not exist and this appears to be a direct reaction to the eugenic excesses of the Nazi regime. In other countries, factors operate relating to the concerns of ethical committees about confidentiality and EU directives. An essential prerequisite is the assurance of absolute confidentiality and the protection of data from third parties. The putative benefits are currently being evaluated by a UK government-funded controlled trial.

## Dimension 6: State of the Art

*Education and Training*

Most authors provided information on the training and educational aspects of medical genetics, although this varied greatly in detail, often mirroring a lack of national organisation (table 7). The importance of education clearly extends beyond genetic specialists because one of the most serious obstacles to access to genetic services is lack of professional education about genetics for physicians practising in primary or secondary care. The importance of clinically relevant genetics education for medical and nursing students is widely appreciated but only a few authors could document formal agreements on teaching details.

The collaborating countries have different levels of sophistication of organisation and delivery of specialist training in medical genetics. However, the general pattern advocated for specialist genetic physicians is similar and consists of a basic medical qualification followed by general clinical training in a range of subjects including paediatrics, general internal medicine and obstetrics and gynaecology. This is followed by a period of specialist training in a genetics centre and generally lasts 4–7 years. After 2 years of specialist genetics training, a certificate of completion of training can be obtained in the EU and this allows entry to a specialist register. Recognition of training centres, with or without inspection, is mandatory only in some countries, as is formal assessment of trainees.

Formal training schemes for laboratory scientists working in genetic services may not yet exist in many countries and in only a few countries are there clear guidelines for their training and accreditation. In some countries, medical graduates are in charge of genetics laboratories but in some, scientists with PhDs or MScs direct laboratories with laboratory technicians engaged in bench work. Genetic co-workers (e.g. genetic nurses, counsellors), where they exist at all, receive only incidental on-the-job training although a few countries have established courses (e.g. UK, Greece).

**Table 7.** Education and training: data provided by CAGSE authors

| Country | Undergraduate | Postgraduate |
|---|---|---|
| Austria | There are no published recommendations for teaching genetics to medical students. | Recent recommendations for training medical geneticists are not expected to be effective as there are no posts or centres for training. |
| Belgium | Inadequate teaching of genetics to medical students, nurses and midwives, poor assessment of training in clinical genetics, little education of primary care staff and non-availability of national guidelines. However, integrated multidisciplinary consultation and counselling services for hereditary cancer syndromes are being set up in different centres. As part of this effort, residents in one of the clinical specialities are offered PhD training in human genetics. | |
| Bulgaria | Genetics first taught to medical students in 1919 when Medical Faculty in Sofia was founded. Third year in the Department of Medical Genetics of the Medical Faculties: 56 hours + 8 hours clinical genetics in the 5th year paediatrics course. | Clinical genetics in 3-month general and short-term specialised paediatric and obstetric courses. Intensive course of molecular biology for teachers. 16 hours for nurses and midwifes in Sofia in the last year of their education. |
| Croatia | In cell biology, paediatrics + 20 hours on ethics in genetics. Four years molecular biology: about 30 students annually likely to be a good source of medical molecular geneticists. Nurses and midwives receive genetics teaching from trained geneticists in nursing school. | Recently postgraduate training in medical genetics is in process of being reorganised, will last four semesters and offer a broad choice of related subjects. Non-clinical and molecular genetics courses organised twice yearly. |
| Cyprus | No medical school. | Training programmes organised locally and abroad by the Ministry of Health and Cyprus Institute of Neurology and Genetics. |
| Czech Republic | No standard level of undergraduate education of medical students in the seven different medical faculties. Quality and quantity of clinical part of medical education differs in each medical school. At some it is minimal or not compulsory. | Systematic postgraduate education in clinical genetics from late 1960s in Prague Since 1980, speciality has expanded. |
| Denmark | Cell biology in the first years of medical school; students at the University of Copenhagen have 30 hours of lectures in human genetics and 25 hours of lectures in clinical genetics. | A postgraduate 1-week course in clinical genetics yearly, attended mostly by specialists in paediatrics or gynaecology/obstetrics. From 1996, new training programme. |
| Finland | Several chairs of medical genetics providing preclinical studies for medical students; medical faculties without departments of medical genetics and the clinical departments have integrated teaching of genetics in their own teaching programmes. | Specialisation programme for medical geneticists run by three medical faculties. Programme run by the University of Helsinki for non-medical 'hospital geneticists' who can run laboratories. No organised special training for genetic nurses. |
| France | In 1st year (approximately 30 hours), sometimes by instructors from the Science Faculty. Eight of the 37 Faculties of Medicine have no professor of medical genetics. | Review boards legally established 1990 and physicians able to qualify in medical genetics. No training in genetics for nurses, midwives and psychologists: profession of genetic counsellors does not exist due to opposition of medical geneticists. |
| Germany | Genetics is taught with basic biological training but time and contents not specified. | Detailed requirements for training for specialists in human genetics and non-medical postgraduates. No training for genetic nurses. |
| Greece | Genetic teaching in medical schools is insufficient. | Geneticists well trained by visiting best foreign centres. Applications for Masters counsellors course greatly exceed positions. Graduate nursing students: practical application counselling. |
| Hungary | Medical genetics education for medical students unsatisfactory: explains inadequate knowledge of medical graduates. | Recognition without examination, unlike majority of specialties; no postgraduate course in medical genetics for 15 years; no status/training for genetic coworkers. |
| Ireland | Basic genetics taught to medical students in all Irish universities but content varies widely. | There are no published recommendations for teaching or training in genetics and would look to the UK and the USA. |
| Israel | Part of preclinical studies in all universities; lectures in paediatrics, gynaecology and haematology and common final medical examination for all students in four universities. | Two-year training programme in approved medical genetics centres leading to board examinations and certification; must be in approved medical genetics centre; included in schools of nursing. |

(Table 7 continued next page.)

**Table 7.** (continued)

| Country | Undergraduate | Postgraduate |
|---|---|---|
| Italy | There are no published recommendations for teaching genetics to medical students. | No published training for medical geneticists, laboratory geneticists, or genetic nurses/associates. However PhD of Postgraduate School of Medical Genetics – 4-year course. |
| Latvia | From 1995, 38 lecture hours and 38 laboratory hours were established in the Department of Medical Biology and Genetics but medical geneticists not directly involved. | No accredited training programmes. |
| Lithuania | From 1990 organised course for medical students. | From 1991 special training programmes for medical geneticists. |
| Netherlands | No data. | Training in clinical genetics is only possible in centres accredited by Royal Dutch Medical Society; cytogeneticists and molecular geneticists have made progress in planning training programmes. |
| Norway | Both human genetics and clinical genetics in all universities. | Approval of satisfactory training by Norwegian Medical Association. |
| Poland | Uniform programme of medical genetics for undergraduate students was implemented 1996 (minimum time for lectures and practice 30 hours). | 1997: specialisation in medical genetics may formally be established. |
| Portugal | Included in the curriculum of all faculties of medicine. | Training programme comprising 5–7 years in addition to 8 years undergraduate and intern. |
| Romania | No published recommendations for teaching genetics to medical students, training medical geneticists, training laboratory geneticists, training genetic nurses/associates. | |
| Russia | Four chairs of medical genetics have recently been established in Moscow, St. Petersburg and Tomsk with special emphasis of medical genetics input to clinical disciplines. | Primary training programmes for clinicians and for scientists overseen by special Department of Medical Genetics of the Russian Medical Academy for Postgraduate Study. |
| Serbia-Montenegro | Undergraduate medical studies include biology and human genetics in the first year with 60 hours of theoretical and 45 hours of practical work. | Two year postgraduate course in human genetics to become Master of Science. |
| Slovenia | Since 1972, medical genetics has been taught at the Institute of Cell Biology, Medical Faculty Ljubljana with participation of Department of Medical Genetics. | Recently developed a training programme for medical geneticist (2 years) as well as for clinical cytogeneticists and clinical molecular geneticists (each lasting 1 year). |
| Spain | Since 1990, medical schools required to include human genetics. | There are no official training programmes in clinical genetics, cytogenetics or molecular genetics. |
| Sweden | National consensus among medical geneticists on what should be taught but still great local variations among the six medical schools. | Specialist training regulated by National Board of Health and Welfare. |
| Switzerland | No national agreement teaching genetics to medical students and nurses and differences between university centres. | No defined training for clinical or laboratory scientists. |
| Turkey | No data. | speciality training in 'medical genetics' but no system to standardise the curriculum; no programme for teaching genetics to primary health care staff, nurses/midwives. |
| Ukraine | A Department of Clinical Genetics and Clinical Immunology established at the Medical Institute of Lugansk for teaching genetics to medical students. | Two departments of medical genetics at the institutes of advanced studies provide training in medical genetics for physicians (in Kiev and Kharkov). |
| United Kingdom | Earlier reviews and recommendations rendered redundant by new General Medical Council problem-based curricula. | Postgraduate training planned and monitored by the Royal Colleges. |

### Dimension 7: Consumer Satisfaction

The importance of public debate about genetics is referred to in many country chapters. Although lively discussion is reported in some, it is not evident that this is true in all, nor is the extent known of public knowledge of the implications of genetic developments. The diffusion of prenatal screening across Europe, including the influence of consumers has been studied [3]. However the acceptability of the services has not been studied in many countries, and indeed only in a handful (e.g. Germany [8], Poland [9]) has research investigated the attitudes of women to the services (usually prenatal screening). This work has led to greater care to explain the purpose of routine screening, such as maternal serum screening.

*Consumer Organisations*

The majority of country chapters reported that there was consumer involvement in the services, often through the work of patient organisations and societies. Indeed, authors report a wide range of patient organisations, although their scope is not always clear, nor where the boundaries of their work lie. 'Umbrella' consumer organisations representing many individual disease-specific groups have become influential in the Netherlands, UK and several other European countries and with their concerted voices exert increasing influence on governments.

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