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Genetic Services in Denmark

Country Background: Demography, Geography and Infrastructure

Denmark consists of the Jutland peninsula, 4 main islands and approximately 480 small islands, with the area totalling 43,093 km^2. It is a parliamentary democracy with a constitutional monarchy and has a population of approximately 5,187,000, with 122 people/km^2; 86% of the population live in urban areas and approximately 25% live in Copenhagen; 90.6% of the population are Lutheran Protestant. Denmark has a good transport and communications network and has a 100% literacy rate. It is very homogenous, 96.2% of the population are Danish with one ethnic community of German speakers, who live on the border with Germany in Jutland. Foreign nationals constitute 3.8% of the total population and include citizens from Sweden, Norway, Finland, EU countries, Africa, Asia and South America. There are 20,000 refugees from the former Yugoslavia. The living standard is among the highest in Europe, as are the tax rates that are necessary to pay for the welfare system.

Health Service Setting

Expenditure on health care is approximately 6.5% of the GDP. The overall responsibility for health care lies with the Ministry of Health. The National Board of Health has an important role in suggesting guidelines, coordinating and advising the minister. The management of health care facilities is largely decentralised, and the system is about 85% financed by local and regional taxes. The remaining approximately 15% is paid by the patients themselves. The health insurance system covers 100% of the population, and almost 95% of the population has chosen the group one scheme, which implies access, almost free of cost, to comprehensive health care, including free medical care from the GP and free specialist care after referral; 5% have chosen the group two scheme with freedom to consult any doctor of their choice, but they are charged 50% of the costs of out-patient care. Hospital in-patient care is free of charge to both groups. Most specialist care is provided by hospital-based salaried medical specialists. Only a few hospitals are private and there is no private general practice in Denmark.

There are 28 doctors and 64 hospital beds per 10,000 people. In 1990 there were 14,277 physicians in Denmark, or an average of 1 per 360 citizens. Among them were 3,208 GPs who play an important role in preventative medicine: carrying out antenatal care examinations, providing postnatal care and care of preschool children. According to the Nivel Report in 1990, there was 1 GP per 1,609 inhabitants. Provision of primary information and counselling to pregnant women is by GPs, who can refer the women to genetic departments for further investigations and counselling.

History of Medical Genetics

Clinical genetics has evolved from the university institutes of human genetics in Copenhagen, Aarhus and Odense, as well as within the departments of paediatrics and gynaecology/obstetrics at Rigshospitalet, Copenha-

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Table 1. Medical genetics staff in genetic service at the J.F. Kennedy Centre, Glostrup

Centre name	Population served	Genetically trained physicians	Genetic physicians in training	Cytogeneticists + technicians	Molecular geneticists/ technicians	Genetic nurses/ counsellors
JFK	500,000	4	2	15	6	0

Table 2. Medical genetics staff engaged in genetic service provision in Denmark

Number of centres	Population served	Genetically trained physicians	Genetic physicians in training	Other staff
6	5.1 million	about 20	about 5	130

gen. Research into the genetics of mental retardation is a particular strength of the John F. Kennedy Institute, Glostrup. Clinical genetics has very recently been recognised as a separate medical speciality in Denmark (1996).

Dimension 1: Availability

Genetics was only recently (1996) recognised as a medical speciality officially comparable to paediatrics (for example). Currently, there are government plans being implemented to expand genetic services in Denmark. Six genetic departments in Denmark provide genetic counselling and variable amounts of laboratory investigations. Around 20 people at academic level are involved in genetic counselling and laboratory testing, but these numbers are imprecise, as some clinical genetics work is done part-time by researchers. At the moment there are five laboratories doing cytogenetic prenatal analyses (one has recently started in Odense). Tables 1 and 2 show the medical genetics staff in the author's centre and in Denmark as a whole.

Not all counselling is specialised, some counselling is done in paediatric and gynaecological departments by doctors and midwives, and advice by telephone from geneticists to assist counselling by colleagues is well developed.

Prenatal cytogenetic diagnosis was first performed in Denmark in 1970, and after eight analyses during the first year the number has now increased to around 8,500 analyses per year. This means that around 12% of all pregnancies are investigated by invasive prenatal procedures. The majority are done for risk of a chromosomal disorder, about 5% for risk of a neural tube defect, and approximately 1% for a monogenic disorder. The proportion of analyses performed on chorionic villus samples (CVS) has increased substantially in recent years and now constitutes nearly 40% of all prenatal diagnoses. The age limit for indication through advanced maternal age is 35 years. Acceptance of prenatal diagnosis is widespread: about 65–70% of pregnant women over 35 have an amniocentesis or CVS. Urban areas have the highest uptake.

Maternal serum AFP screening and combinations with other serum markers with the aim of identifying both neural tube defects and Down syndrome was carried out as a project in certain centres. Around 20% of pregnant women were screened for high AFP in the years 1980–1991, and for low AFP from 1991 to 1994. General screening is not recommended by health authorities (National Board of Health) at the moment. In Denmark only one small region (3,000 pregnancies per year) has triple testing.

General ultrasound screening is similarly not recommended by authorities. Nevertheless, around 76% of women have an ultrasound investigation during pregnancy.

A few analyses have been centralised to one centre. This applies to cystic fibrosis (CF) testing, which is done at Rigshospitalet, Huntington testing at the Department of Medical Genetics, University of Copenhagen, and PKU and Menkes syndrome mutation analyses at the John F. Kennedy Institute. A number of analyses are done within the context of research programmes, and for that reason are linked to one laboratory. At the moment, a council within the framework of the Danish Society of

Medical Genetics is exploring the possibilities for mutual agreement to centralisation of analyses and counselling of specific diseases.

Newborn screening for PKU and hypothyroidism is nationwide. A screening programme for 21-hydroxylase deficiency is starting as a pilot study. All PKU children are treated at the J.F. Kennedy Institute. Treatment of hypothyroidism has been decentralised to regional paediatric departments. Treatment of CF is done in two centres. Newborn screening for CF has been discussed, but so far the opinion is against. Health authorities have recommended centralisation of medical treatment of rare congenital handicaps and diseases. CF carrier screening was carried out as a pilot project at Rigshospitalet in the years 1990–1992, screening approximately 7,000 pregnant women. One CF fetus was identified and aborted.

Genetic Registers

A national congenital malformation registry reports regularly. All induced abortions are registered in a separate register. A cytogenetic central registry contains all details of cytogenetic analyses, both prenatal and postnatal. Annual reports summarise the findings, from which the number of abnormalities found in prenatal investigations can be seen in relation to e.g. indication groups, maternal age and region. A register for all FAP families exists, as well as for some other inherited tumour syndromes (Von Hippel-Lindau, neurofibromatosis, retinoblastoma).

Dimension 2: Access

In principle, all genetic services and testing are free of charge for patients and families. There are so far no private laboratories performing genetic testing. However, GPs cannot at the moment request genetic tests directly for their patients, as the request must come from a specialised hospital department (for instance, paediatric or obstetric) to be funded by the health care insurance system.

Factors impeding access to genetic services in Denmark are:

(1) administrators poorly informed about genetics;
(2) poor reimbursement for genetic analysis;
(3) poor co-operation between geneticists and general practitioners;
(4) lack of direct access to specialised analysis for general practitioners;
(5) too few genetic specialists;
(6) difficulty in funding new posts for genetically trained physicians and specialist laboratory geneticists;
(7) difficulty in funding new tests;
(8) lack of continuity of care for patients/families with long-term genetic problems, and
(9) poor educational level of patients concerning genetics and poor public awareness of genetic services.

Poor co-ordination amongst laboratories and amongst clinical geneticists, poor networking amongst centres for rare disorders and poor organisation in public hospitals/centres are a result of the, until 1996, lack of formal recognition of clinical genetics services. Collaboration is therefore dependent upon personal relations. Hopefully, a number of these problems will be amended with the recognition of clinical genetics. Access to genetically trained physicians, trained genetic laboratory staff and clinical molecular genetic services is at the moment good only in some areas of the country but this is improving. Specialist genetic nurses/counsellors, population screening for carriers of recessive disorders and general genetic registers, apart from some research registers, are non-existent. However, access to newborn screening and cytogenetic registers is excellent everywhere.

Free (self-determined) abortion up to end of the 12th week of pregnancy has been legal in Denmark since 1973. After the 12th week, a local council has to grant permission on social or medical grounds. Prenatal diagnosis of a serious handicap in the fetus in the second trimester is an acceptable indication for induced abortion if the couple so wishes. There is in principle no legal limit to gestational age for induced abortion, but in practice 24 weeks is a limit which is rarely exceeded, and then only for lethal disorders. The Danish National Board of Health sets written guidelines and recommendations for indications for prenatal diagnosis; the latest was published in December 1994. The basis for these guidelines had been accepted by the Folketinget as a part of pregnancy care.

Dimension 3: Life Sustaining

The annual birth rate has increased in recent years, approaching 70,000 newborns per year, after a minimum of around 50,000 in the early 1980s. The fertility rate is now 1.68 children per woman. Infant mortality is 7.5 per thousand live births. Life expectancy is 72 years (men), 77 years (women). Maternal age at childbirth has increased in the last 25 years and mean maternal age is now 28

years, so that more children are now born to mothers aged 30–34 years than to mothers aged 20–24 years.

At the moment, very little testing for cancer susceptibility genes is carried out, and so far only in individual at-risk families. Cancer family counselling clinics within clinical genetics are under development. A Nordic study in collaboration with the National Cancer Institute is planned for all known ataxia teleangiectasia (AT) families to evaluate the risk of breast cancer in ATM carriers.

Dimension 4: State of the Art

Formal rules for training to qualify as a medical/clinical geneticist have been formulated and in principle are valid from August 1996, but in practice there will be an interregnum. After basic training for 18 months (common to all specialities), a 6-year postgraduate training programme will contain elements of formal genetics, laboratory work, including prenatal and molecular genetic analyses, as well as education in counselling and a general clinical training in either paediatrics, gynaecology/obstetrics, or internal medicine/neurology. Thirty-six months are spent primarily in clinical training, including counselling, and a further 36 months will encompass cytogenetic and molecular genetic laboratory work.

With regard to teaching genetics to medical students, training in genetics for physicians, molecular technology, up-to-date laboratory equipment and co-operation with other countries is good only in some areas. Teaching of genetics to medical students: in addition to cell biology in the first years of medical school, students at the University of Copenhagen have 30 h of lectures in human genetics and 25 h of lectures in clinical genetics. Revisions planned include an increase in the teaching of genetics in medical school.

A 1-week postgraduate course in clinical genetics is arranged yearly under the auspices of the National Board of Health, attended mostly by specialists in paediatrics or gynaecology/obstetrics. It is planned to expand this course into an interdisciplinary intensive training course aiming at both clinical geneticists in training and other specialists (e.g. neurology, internal medicine).

Dimension 5: Non-Harmful

All health care professionals including clinical geneticists are supervised by the National Board of Health and can be subject to sanctions for poor practices. All laboratories have developed some form of quality control programme, but none are ISO 9,000 certified. Quality assurance programmes for prenatal diagnosis and for counselling in Huntington disease are under development.

Apart from this, a National Ethical Council is very active in initiating discussions about ethical questions arising from the new genetics, especially in relation to ethical values, community interests and individuals' integrity. Questions of genetic registers, prenatal diagnosis, gene therapy and genetic screening have been discussed, and reports are available.

Filing of patient data is regulated by law. Patient files are strictly confidential, and the patient must give informed consent for anybody to take part of the information.

Although there is currently a lack of quality assurance in laboratory cytogenetic and molecular genetics, such assurance is under development. At the moment there is little legislation regulating genetic testing and gene therapy, but the minister of health is expected to regulate by law, for instance, IVF, investigations of eggs and embryos, preimplantation testing and gene therapy.

Dimension 6: Effectiveness

It is difficult to define effectiveness in the context of achievements in clinical genetics and there is caution about cost-benefit analyses which easily develop a tinge of eugenic thinking. There is general agreement that the services provided and counselling should be non-directive. The aims of prenatal counselling have been suggested by the National Board of Health as (1) ensuring that the pregnant women with an increased risk for a diseased/handicapped child is offered relevant services (options) to allow well-informed decisions in the concrete situation, and (2) ensuring that counselling and testing are done with respect for the woman's autonomy and integrity, as well as respecting the fetus.

As a result of prenatal diagnosis the incidence of children with Down syndrome born to women over 35 years has decreased substantially. However, a total decrease in Down syndrome birth incidence over the past 25 years has not been observed in Denmark. This could be due to several factors working together, of which improved registration and especially an increase in mean maternal age are probably the main elements. The county of Funen with approximately 500,000 inhabitants is part of the EUROCAT surveillance of congenital malformations.

The option of prenatal diagnosis in high-risk families (for instance, cystic fibrosis and spinal muscular atrophy) has undoubtedly encouraged couples to start pregnancies they might otherwise have avoided.

Dimension 7: Consumer Satisfaction

There are few investigations of the social impact of genetic services. Tabor and Jonsson published a study of the psychological impact of amniocentesis on low-risk women. An interview investigation of the psychological impact of cystic fibrosis carrier screening has been published.

Danes are fond of organisations, and a large number of patient and parent organisations exist, both very large covering a broad range of handicaps, and very small ones for specific rare diseases. A 'centre for rare handicaps', was established by the Ministry of Social Affairs, and here all types of professionals and individual families can obtain information and advice about e.g. experts, resource people and social rights concerned with specific handicaps.

The activities of genetic interest groups and the public awareness of, and responsiveness to, genetic services are very important to the present situation and development of genetic services in Denmark.

Publications Related to Genetic Services

1 Yearly Report from Cytogenetic Central Register in Denmark.
2 Report from the National Board of Health in Congenital Malformations.
3 EUROCAT reports.
4 Fostervandsundersøgelser og moderkagebiopsier 1990. Cytogenetisk Central register og Sunhedsstyrelsen. Vitalstatistik 1:32: 1992.
5 Medicinsk fødsels-og misdannelsesstatistik 1991. Sundhedsstyrelsen Vitalstatistik 1:35:1993.

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'It has been validated by me and is approved by the Danish Society of Medical Genetics.' This chapter was also circulated to all clinical genetics departments as well as to the Minister of Health and the National Board of Health.