European Journal of Human Genetics

Eur J Hum Genet 1997;5(suppl 2):130–134

Genetic Services in Norway

L. Tranebjærg
with the co-operation of *Anne-Lise Børresen-Dale, Inger-Lise Hansteen, Sverre Heim, Eli Anne Kvittingen, and Pål Møller*

Department of Medical Genetics,
Regionsykehuseti Tromsø, Norway

Country Background: Demography and Geography

Norway is a large country from north to south but has a population of only 4.3 million and a population density of 14/km^2. Approximately 11% of the population live in Oslo and 63% live in urban areas of more than 2,000 people; 95% of the population are members of the Church of Norway. The population is very stable, 3.4% are foreign citizens coming from other European countries, as well as Africa and Pakistan (approximately 1%). One-third of all foreign citizens, in particular from Pakistan, live in Oslo. Norway has a good transport infrastructure and living standards are high: 20% of adults have completed higher education (university or similar), 98% of the population have a television set, and 39% have access to a personal computer at home. In Norway, 4.9% are unemployed. Norway voted against membership of the EU in 1994.

Type of Health Service

Expenditure on health care in 1973 was approximately 7.6% of GDP. General health care, policy-making and legislation are the domain of the Ministry of Health and Social Affairs, which also has the ultimate responsibility for financing and planning health care. Other important central functions are within the Directory of Health Services and the National Insurance Institute. The Directorate consists of several departments: one for primary health care, one for hospital care, one for provision of drugs and one for environmental health as well as one for administrative activities.

The total number of physicians in 1996 was 15,368. There are on average 35 hospital beds and 34 doctors per 10,000 people. A compulsory national insurance scheme covers the whole population. Health care is financed from three sources: premiums paid by employees and self-employed persons, contributions by employers and contributions by the state, counties and municipalities. The insurance scheme ensures that in-patient treatment in public hospitals is free of charge. Out-patient consultations with specialists are subject to cost sharing.

Planning and management of hospitals and specialised health services are the responsibility of the 19 counties. Hospitals can be categorised according to a hierarchical structure of regional, county and district hospitals. Each of the five health regions into which Norway is subdivided has its own regional hospital owned by the county in which it is located. Only the national hospital in the region of Oslo, which is highly specialised, is owned by the state.

Planning, funding and organisation of primary health care and prevention is the responsibility of 450 municipalities. Four types of service are compulsory: general practice, physiotherapy, public health nursing and home nursing. The role of the municipalities implies being a contractor and an employer of health providers. Primary health care is now largely decentralised following the Municipal Health Act of 1984.

History of Medical Genetics

Clinical genetics has evolved from university institutes of medical genetics, first in Oslo, later in Bergen. A third department of medical genetics was started in 1986 in the University Hospital of Tromsø. The three departments of medical genetics cover the majority of postnatal cytogenetic services, and prenatal diagnosis to all five health

KARGER

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

L. Tranebjærg
Department of Medical Genetics
Regionsykehuset i Tromsø
N–9038 Tromsø (Norway)

regions. In addition, a smaller cytogenetic laboratory is associated with a Research Laboratory for Occupational and Environmental Genetics at a district hospital in Skien, southern Norway.

Dimension 1: Availability

By the end of 1993, there were 22 approved specialists in medical genetics, but only 11 of those are working full-time in medical genetics. In 1993, there were 7 training positions; in 1995 the number was increased to 10. In total there are 18 positions for MD fully trained specialists in medical genetics, of which 3 are presently vacant.

Neonatal screening for PKU has been available since 1967 and from 1978 screening covered nationally all newborns. Based on the number of abnormal newborns in that period, the incidence in Norway of PKU has been estimated be to 1:13,250. Recently, the mutational spectrum of PKU has been published and mutation characterisation of PKU cases is concentrated in the Department of Medical Genetics in Bergen. Neonatal screening for hypothyroidism started in 1979 and based on positive diagnoses during 1979–1993, the incidence is estimated at 1:3,200. Newborn screening for other diseases is currently not available, although screening has been discussed for galactosaemia, 21α-hydroxylase deficiency and cystic fibrosis. In Norway, only 60% of cystic fibrosis patients have ΔF508. The Biotechnology Act (see below) states the parents' right to 'require' newborn screening if early diagnosis of a disorder significantly improves the prognosis.

Most diagnoses for disorders of metabolism are performed by the Department of Clinical Chemistry at the National University Hospital in Oslo. In 1995, approximately 1,200 analyses were performed and the yield of abnormal samples was estimated at 2% (no formal statistics exist). Specialised oximetry studies of fresh muscle samples, assays of the respiratory chain enzymes, and investigations of mitochondrial mutations are available at the same hospital.

Representatives from the departments of medical genetics meet once a year. In Norway there is no handbook describing the availability of genetic and biochemical studies in the various laboratories. Based on long-term research involvement, biochemical and molecular investigations for tyrosinaemia, type 1, are done at the National University Hospital of Oslo. Presymptomatic counselling and testing for Huntington disease is performed only in the Department of Medical Genetics, Ullevål Hospital, Oslo. In 1989, a service for genetic oncology was established at the Norwegian Radium Hospital in Oslo, and since 1995, resources have been given to the hospital for genetic counselling of cancer families. The department of genetics has a research-based long-term experience both in cytogenetics and in molecular biology and mutation screening.

Involvement of specially trained nurses, with a subsequent university degree, is not generally accepted as part of a medical genetic centre, but at the present time there are three such positions in Norway in two different genetic departments.

Dimension 2: Access

Initial information for pregnant women is given by GPs who can refer them to genetic departments for genetic counselling if necessary. Preventive health investigations of infants are also primarily covered by GPs who can refer to paediatricians if needed.

Until 1995, financial resources to genetic services, especially prenatal studies, were by block grants earmarked for these activities in three centres. From 1995, financial resources for genetic services were combined with other resources into one general block grant to the county hospital. Resources for genetic counselling are given by the municipality of Oslo to the Department of Medical Genetics, Ullevål Hospital. In addition to this, the Radium Hospital, also in Oslo, receives financial support for genetic counselling of cancer families. Otherwise, no resources are given by the state, or the counties elsewhere in Norway, for genetic counselling.

There is a scheme of coverage of partial expenses of postnatal cytogenetic analysis and out-patient genetic counselling from the patient's municipality and county, and from January 1, 1997, there will be partial coverage of expenses for molecular analysis. In the present situation, laboratory genetic services cannot be based on income from analyses performed.

Criteria for prenatal diagnosis were circulated in 1983 in a recommendation by the Directory of Health. These included advanced maternal age (above 38 years at term) and high-risk groups where a Mendelian disorder or a chromosome abnormality has been diagnosed. In Norway, all pregnant women are offered ultrasound examination at the local hospital during weeks 17–18. Recently, information has been published by the Directory of Health to alert pregnant women to the fact that such ultrasound examination may reveal serious abnormalities in the fetus, and to reassure them that the examination is

voluntary. The majority of females eligible for prenatal (invasive) studies are informed by a medical geneticist, but due to the absence of a medical genetic department in one large health region around Trondheim, the information there is given by a specialist in gynaecology in the district department of obstetrics and gynaecology. Amniocentesis is usually performed during weeks 13–15, with regional variations. In 1993, 1,143 amniocenteses were carried out. Chorionic villus sampling (CVS) is available only in Trondheim and Oslo centres, is rarely used and then almost exclusively for high-risk pregnancies; in 1993 only 44 CVS were performed. In 69%, the indication for amniocentesis was advanced maternal age ($\geq$38 years at expected term). Close to 14% of the amniocenteses were performed after demonstration of abnormalities at ultrasound examination; 28% of the amniocenteses in this group of patients showed chromosome abnormalities. The number of live births in 1993 was 60,100 and the total number of prenatal studies in the same year was 1,187. This means that 3.0% of all live births had a prenatal study. In 1993 and previous years there was a significant decrease in uptake of prenatal diagnosis from south to north, from approximately 60% in Oslo to 29% in the northernmost county in Norway (Finnmark). In one particular western county, the uptake rate is approximately 15%, while the average national uptake of prenatal diagnosis for the indication advanced maternal age is around 40%. Deliveries outside hospitals amount to 1.7%.

Dimension 3: Life Sustaining

The Medical Birth Registry of Norway, at the University of Bergen, compiles data from the compulsory registration of deliveries. The annual birthrate was 60,100 in 1993. The fertility rate is now 1.87 children per women. Infant mortality is 5.2 per 1,000 live births. Life expectancy for women is 80 years and for men 75 years. Mean maternal age at delivery is 28 years, and 11.2% of mothers are $\geq$35 years at the birth of their child.

Dimension 4: State of the Art

Medical genetics has been a recognised medical speciality in Norway since 1971. Medical students are taught both human genetics and clinical genetics late in medical school in all universities. So far the total number of lectures within medical schools is approximately 40. As part of the formalised training programme in medical genetics, every second year a 21-hour course in cancer genetics is given.

Training in medical genetics is formalised and lasts for 5 years, including one compulsory year in some other clinical speciality. A committee in each department supervises training and refers to a national board in medical genetics set up by the Norwegian Medical Association. The goals and scope of training are being re-evaluated but at the present time these include a requirement of 1 full year training in a molecular laboratory, 1 year cytogenetic training and 2 years training in genetic counselling covering a variety of genetic disorders and prenatal counselling. In addition there is a requirement to attend 120 h of theoretical teaching. The approval of satisfactory training is given by the national Board of Medical Genetics. No formal exam needs to be passed.

Postgraduate medical genetics courses for GPs and other clinical specialists are very limited.

Dimension 5: Non-Harmful

Legislation Relevant for Genetic Services

Free self-determined abortion is legal in Norway until the end of week 12. In 1994, 14,533 abortions were performed, corresponding to 24.2% of the live-born rate. Abortion after the end of week 12 must be approved by a local council when it can be permitted on social and medical grounds, including prenatal diagnosis of a serious handicap. Termination of pregnancy after week 18 can only be permitted if the condition in the fetus is lethal.

In August 1994, the 'Act relating to the application of biotechnology in medicine' (Act No. 56 of August 5, 1994) was passed by the Norwegian Parliament. The Act gives a frame of general guidelines for assisted reproductive technology applications, research on embryos, preimplantation diagnosis, prenatal diagnosis, genetic testing after birth and gene therapy. In addition, the Act also specifies obligations about authorisation of institutions applying medical biotechnology and the duty for such institutions to report regularly on their activities to the Ministry of Health and Social Affairs, which has passed a consultative frame on to the Directory of Health Services specifying how the legislation shall be implemented. The Directory has delegated the implementation of parts of the Act to the National Health Board which has nominated a 'Medical Technology Advisory Board' consisting of ten specialists (MDs or PhDs) with long-standing experience in applying medical biotechnology.

Genetic testing for diagnostic purposes is permitted without restrictions. However, the law requires that comprehensive genetic counselling be given before, during and after genetic tests performed on healthy persons for presymptomatic, predictive or carrier purposes. Presymptomatic, predictive and carrier testing is limited to individuals above the age of 16 years. When the information refers to a diagnostic test, genetic results may be communicated, without restrictions, between medical institutions authorised to apply medical biotechnology. However, the exchange of genetic information about presymptomatic, predictive or carrier tests is restricted. Notably, the Act states that it is even prohibited to ask (i.e. as an insurance company or an employer) whether a presymptomatic/predictive/carrier test has been performed. Gene therapy is only allowed as somatic cell therapy, and minors (below 16 years) need the consent of their parents/guardians. The Act is to be reviewed in 1999.

An Advisory Board of Biotechnology is a broadly based board of representatives giving advice to the government on questions relating to genetics and ethical issues. It is not restricted to medical biotechnology. The Advisory Board has a consultant function for general and ethical questions around genetic tests, and application of biotechnology not restricted to the medical area.

Health care provision is supervised by the Directory of Health Services. As part of the implementation of the Biotechnology Act, criteria are being set for the requirements within each medical genetics centre to assure properly qualified services. No laboratories of medical genetics in Norway are accredited but the goal of the Biotechnology Act is to assure that all laboratories fulfil good laboratory practice, and quality assurance programmes for various laboratory studies (cytogenetics and molecular genetics) are being established. The Biotechnology Advisory Board promotes discussion of ethical aspects of genetics, arranges open meetings for the public and publishes a journal (*Gen Etikk*).

No national cytogenetic or prenatal cytogenetic registry is available. In the period 1989–1993, prenatal activity was compiled in a report by the Directory of Health (Eterprøvningsutvalget, 1989–1993) and a new way of reporting is being developed. The medical Birth Registry of Norway publishes an annual report which is widely available. Strict regulations applying to patient data are administered by 'Datatilsynet', and the rules to secure a person's confidentiality are very strict.

Dimension 6: Effectiveness

Prenatal diagnosis is available for women at increased risk. The nationwide availability of ultrasound examination provides accuracy of gestational age and detects possible complications of pregnancy. Of all abortions in the period 1979–1994, 98% were before the end of week 12. Among women in the age group 15–49 years, 13.5 per 1,000 terminated pregnancies. Among the 1,316 amniocenteses performed in 1993, 6.2% showed abnormality and 4.2% women chose termination of the pregnancy based on demonstrated abnormality. Among patients with abnormal prenatal findings, 68% (56/82) chose termination of pregnancy. There is no national registration of prenatal diagnosis, or national guidelines for follow-up after prenatal diagnosis. In Norway, there is no maternal serum AFP screening. In the period 1989–1994, a report was compiled each year to provide key figures about the provision of medical genetic services in Norway.

The first report of effectiveness in follow-up for inherited breast and ovarian cancers was published in 1996. A register for autosomal dominant polyposis families exists within the Cancer Registry, and routines are being developed to provide information to all at risk persons about clinical follow-up and genetic counselling.

Dimension 7: Consumer Satisfaction

There is now extensive debate about whether the diagnosis of trisomy 21 should be an indication for termination of the pregnancy, or whether the goal of genetic counselling and prenatal diagnosis is to provide the individual family with the means to plan their family within the framework of risks they may have. The possibility of early prenatal diagnosis in families with a high risk for an inherited disorder has encouraged more families to undertake additional pregnancies after the birth of a handicapped child.

The Department for Social Affairs has initiated several project-based strategies for distributing information about rare genetic disorders by concentrating non-medical experience about ten different rare genetic disorders in an information centre at the University Hospital in Oslo (Smågruppesenteret), and by establishing for a trial period a competence centre providing medical and physiotherapy treatment for patients with Marfan syndrome, arthrogryposis and neural tube defects (Sunnaas Sykehus). Other networks for providing detailed medical and genetic information about rare disorders such as Usher Syn-

dromes, with involvement of the consumers, are under consideration. A large number of patient and family organisations, supporting specific groups of genetic disorders, exist. Several patient organisations have, in collaboration with specialists in medical genetics, produced information material. The Centre of Frambu (Frambu helsesenter) provides regular courses for patients plus their families with particularly rare disorders such as neuromuscular disorders, Usher syndrome, fragile X and others, and provides a strong baseline for well-informed consumers. Specialists in medical genetics serve informally as consultants for a large number of patient organisations. There is strong public awareness of genetic issues, in particular with respect to indications for termination of pregnancy, screening procedures for genetic diseases, and in vitro fertilisation and preimplantation diagnosis.

Acknowledgements

I am deeply indebted to all my colleagues who have helped to compile the data to make this report possible. Special thanks to Pål Møller, Anne-Lise Børresen-Dale and Sverre Heim [The Norwegian Radiumhospital (Cancer Hospital), Montebello, N–0310 Oslo, Norway], Inger-Lise Hansteen (Genetic Section, Telemark Central Hospital, N–3710 Skien, Norway) and Eli Anne Kvittingen (Department of Clinical Chemistry, Rikshospitalet, Pilestredet 32, N–0027 Oslo, Norway).

Publications Related to Genetic Services

1 Official Statistics of Norway. Statistisk Årbok, 1996.
2 Boerma WGW, Jong FAUM de, Mulder PH: Health Care and General Practice across Europe. Utrecht, Netherlands Institute of Primary Health Care (NIVEL), 1993.
3 Etterprøvingsutvalget: Rapport om de medisinske genetiske service funskjoner 1989, 1990, 1991, 1992, 1993 (in Norwegian).
4 Eiklid K, Tranebjærg L, Eiken HG, Pedersen JC, Michaelsen H, Fluge G, Schwartz M, Nielsen B, Bolle R, Skyberg O, Boman H, Berg K: Frequency of the ΔF508 and exon 11 mutations in Norwegian cystic fibrosis patients. Clin Genet 1993;44:12–14.
5 Brøndum-Nielsen K, Nørgaard-Pedersen B: Prenatal diagnostic investigations in the Nordic countries (in Scandinavian with an English summary). Nord Med 1993;108:189–192
6 Medical Birth Registry of Norway (English summary). Annual report 1993.
7 Pettersen RD, Saugstad OD, Heyerdahl S, Motzfeldt K, Lie SO: Neonatal screening for metabolic disorders in Norway (in Norwegian with a brief English summary). Tidsskr Nor Lægeforen 1995; 5:584–587.
8 Pettersen RD, Saugstad OD, Lie SO: Is neonatal screening in Norway on the health political agenda? (in Norwegian with a brief English summary). Tidsskr Nor Lægeforen 1995;5:607–608.
9 Report no. 9 from the Committee for Evaluation of Medical Technology: Consensus conference report about the use of ultrasound in pregnancy. Norwegian Research Council, 1995.
10 Cancer in Norway 1992. Report on cancer incidence and cancer patient survival, 1995 (in English).
11 Act No 56 of August 5, 1994: The act relating to the application of biotechnology in medicine. Ministry of Health and Social Affairs, Norway, 1995.
12 Eiken HG et al: Relative frequency, heterogenity and geographic clustering of PKU mutations in Norway. Eur J Hum Genet 1996; 4:205–213.
13 Stortingsmelding No 16, 1995–1996: Om erfaring med lov om svangerskapsavbrudd.
14 Annual Reports, 1994, from Haukeland Hospital and Ullevaal Hospital.
15 Møller P, Mæhle L, Heimdal K, Dørum A, Tretli S, Helgerud P, Quist H, Bjørndal H, Kåresen R, Nysted A, Varhaug J, Fjøsne H, Guleng R, Due J, Bøhler P, Giercksky K, Tropé C, Kvinnsland S: Inherited breast carcinoma – Prospective findings in 1194 women at risk. Acta Oncol 1996;35(suppl 8):7–11.
16 Dørum A, Kristensen GB, Abeler V, Tropé C, Møller P: Early detection of familial ovarian cancer. Eur J Cancer 1996;32A: 1645–1651.
17 Myklebost O: First for biotech. Nature 1996;384:208.

Norwegian Association for Medical Genetics

(subgroup of the Norwegian Association for Medical Doctors)

Trond Leren MD, PhD, Chairman
Institute of Medical Genetics
University of Oslo
Ulleval Hospital
Oslo (Norway)
Tel. 47 22 11 98 60
Fax 47 22 11 98 99

Validator

Norwegian Association for Medical Genetics
Trond Leren MD, PhD, Chairman
Institute of Medical Genetics
University of Oslo
Ulleval Hospital
Oslo (Norway)
Tel. 47 22 11 98 60
Fax 47 22 11 98 99

'We approve of this as a reasonable description of the situation for medical genetics services in Norway.'