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Genetic Services in Finland

Country Background: Demography, Geography and Infrastructure

Finland is 338,145 km^2 in size and has a population of 5.1 million. The population density is 15 people/km^2 and 64.3% of the population live in urban areas. The smallest town (Kaskinen) has 1,650 inhabitants and the largest (Helsinki) 557,340. Two-thirds of the population live in the southern part of the country; the north is very sparsely inhabited. Finland is a parliamentary republic with highly developed communications and infrastructure. It is economically well developed with a 100% literacy rate. Forestry, metal industry and engineering are the main economic activities. The official languages are Finnish and Swedish (the latter is the language of 5–6% of the population); 85.9% of the population belong to the Lutheran National Church and 1.1% to the Greek Orthodox Church while 12% of the population are not members of any religious association. Foreign nationals make up 1.2% of the population. The extremely restrictive immigration policy is, however, changing and the number of foreigners will probably increase rapidly in the near future.

Health Service Setting

Finland's central government guides the nationwide development and implementation of social security and health care. Local authorities (455 municipalities in 1995) are responsible for the actual provision of social and health care services in their communities. The services are funded by local tax revenues and by state aid for this purpose. Private health care complements the public health care network [1].

The Ministry of Social Affairs and Health [2] is the country's leading authority on social security and health care. The Ministry proposes the main guidelines for social and health policy, designs major reforms and steers their implementation and harmonisation with existing schemes, and serves as a link to the rest of the political decision-making process.

Primary Care

Municipalities must provide local residents with health care services for which a nominal fee is charged. Health care centres usually maintain a number of health care stations in different parts of the municipality or joint authority area. Almost all health care centres have their own small hospitals. The statutory obligations of the health care centres are to provide maternity and child care, school and student health care, and occupational health care. Local authorities are responsible for arranging specialised medical care for their residents. Service above primary level is provided at regional hospitals, central hospitals and the five university hospitals. Health care centres refer patients to a hospital capable of providing the specific type of care needed. There are 20 doctors and 135 hospital beds per 10,000 people.

The whole population is covered by the comprehensive health insurance scheme that includes curative and preventive out-patient care as well as hospital care. Many local authorities produce some of their health care ser-

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Table 1. Medical genetic staff engaged in genetic service provision in Finland

Number of centres	Population served	Medical geneticists (physicians) competent/in training	Hospital geneticists (natural scientists) competent/in training	Genetic nurses	Genetic technicians
8	5,100,000	17/10	9/6	23	60

vices from the private sector. Furthermore, national health insurance reimburses the patient for part of the cost of using private health care services. Finland has a comprehensive and efficient public health care network and only 5% of physicians are engaged solely on private practice. On the other hand, 47% of the doctors employed by the public system have private practice as well.

History of Medical Genetics

Interest in medical genetics increased strongly in the 1960s when the first diseases belonging to the Finnish disease heritage were recognised at Helsinki University Children's Hospital. At the same time, a few cytogenetic laboratories were founded. Genetic counselling started on a small scale in 1952 in Helsinki at the Family Federation of Finland (FFF) in collaboration with the Department of Genetics at the University of Helsinki. FFF is a state-funded union of associations dealing with social, family, and population affairs. Official genetic counselling with the first full-time medical geneticist was started in 1971 at FFF. In 1972, the first chair in medical genetics was founded at the University of Helsinki and the first professor was nominated in 1974. The Department of Medical Genetics at the University of Helsinki was finally established in 1976.

Dimension 1: Availability

Professionals in Medical Genetics (table 1)

Both physicians and biologists have postgraduate university programmes for specialisation in medical genetics (see dimension 4) leading to competent medical geneticists (physicians) or hospital geneticists (biologists), respectively. There are 21 specialists in medical genetics, 17 of whom are employed in the clinical genetics service. The others, mainly involved in research and teaching, occasionally take part in clinical genetic activities. Fifteen hospital geneticists (biologists) are employed in clinical genetics (9 of them are competent and 6 are in the training programme). They run the genetic laboratories. The laboratories can also be run by a medical geneticist or a specialist in clinical chemistry (physicians). There are 18 nurses working full-time in clinical genetics and 7 who work part-time. About 60 technicians are employed for performing chromosome, molecular and biochemical genetic studies.

Clinical Genetics Service – Genetic Counselling

Genetic counselling is a part of the public sector, funded by the municipalities and the state. There are eight genetic counselling clinics; four of them are part of the medical genetics units (tertiary units) located at university hospitals (Helsinki, Tampere, Kuopio and Oulu) and two are separate counselling clinics at the university hospitals in Helsinki and Turku. Furthermore, there are two clinics in Helsinki which serve the whole country: the Department of Medical Genetics at FFF and the Folkhälsan Department of Medical Genetics which began genetic counselling in August 1995, mainly serving the Swedish-speaking population. In the whole country in 1994, there were 3,750 genetic counselling visits altogether, including prenatal diagnosis. No figures are available for genetic counselling for common diseases (e.g. cancer, diabetes) performed by medical specialists other than medical geneticists.

Genetic Laboratories

There are 11 cytogenetic laboratories in Finland, 7 belonging to the public system and 4 private. Two of the public laboratories are at university departments of medical genetics (Helsinki and Turku), four are part of the medical genetics units of the university hospitals and one is part of a university hospital clinical chemistry laboratory. One of the four private laboratories is partly supported by public funds, and is part of an institution for the mentally retarded, the three others are commercial. The annual number of constitutional chromosome studies is about 4,000, prenatal analysis about 5,600 and neoplastic studies 1,800. In 1994, interphase cytogenetics (FISH) and comparative genomic hybridisation (CGH) were performed in 566 cases.

There are 13 molecular genetics laboratories. Molecular genetics is performed at 8 university laboratories. The National Public Health Institute has a molecular genetics laboratory and so has the Finnish Red Cross Blood Transfusion Service. Three private laboratories performing cytogenetics have molecular genetics laboratories as well. About 2,200 samples have been studied per year either with direct or linkage analysis. There were about 600 neoplastic samples; 2,800 samples have been studied for carrier screening for aspartylglucosaminuria (AGU) [3], or coagulation factor 5. HLA studies have been performed for disease association and risk assessment.

The diagnosis of metabolic diseases is performed in five laboratories, all at universities or university hospitals. About 7,000 patients are studied every year for inherited metabolic diseases (organic acids, amino acids, oligosaccharides, glucosaminoglycans).

Genetic Screening

During pregnancy, fetal karyotyping for advanced maternal age or maternal serum screening is offered for e.g. Down syndrome, high α-fetoprotein (AFP), i.e. neural tube defects, and congenital nephrosis of the Finnish type (CNF) [4]. About 65,000 children are born in Finland each year and a rough estimate is that 40,000 mothers participate in the screening.

Ultrasound screening is offered once to all mothers, usually at 18 weeks of pregnancy but in some centres earlier. Abnormal ultrasound findings account for 6% of prenatal chromosome studies and for about half of the abortions for fetal anomalies. The only neonatal screening performed in Finland is for hypothyroidism with almost 100% uptake.

Carrier screening is not routinely done. Pilot programs for AGU, infantile neuronal ceroid lipofuscinosis (INCL) and fragile X are in progress on a limited scale in some parts of Finland only. The tests are offered to women on their first visit to the maternity care centre at 12 weeks of pregnancy. Pretest counselling is given by midwives. Those who turn out to be carriers are referred for genetic counselling arranged by those who run the screening programmes. Accurate figures for participation in these screening programmes are not yet available.

Predictive testing in known families with Huntington disease (HD) is mainly provided by the department of medical genetics at FFF. Some of the other genetic units refer individuals to FFF but some units prefer to carry out the tests themselves. About 10–20 persons request genetic counselling for HD in the whole country annually but very few (0–2) proceed to a predictive test. Based on the experience gained from HD, the department also provides tests for other late-onset neurological diseases, but only a few annually. As part of a scientific research programme, predictive testing is also provided in hereditary non-polyposis colorectal cancer. Studies on other hereditary cancers (breast/ovarian cancer, multiple endocrinological neoplasms) have just started.

Table 2. Figures for genetic counselling, laboratory activity and screening in 1994

Genetic counselling clinics/units	8
Visits	3,750
Cytogenetic laboratories	11
Samples	
Constitutional	4,033
Prenatal	5,621
Neoplastic	1,842
FISH and CGH	566
Total number of samples	12,062
Molecular genetics laboratories	13
Samples	
Direct and linkage	2,259
Neoplastic	585
Screening	2,800
Total number of samples	5,644
Biochemical genetics laboratories	5
Total number of samples	7,000

The figures shown in table 2 have been about the same for several years and there is no reason to expect major fluctuation, except perhaps for carrier screening (fragile X, AGU, INCL).

Long-Term Care Facilities

Long-term follow up is not systematically organised. Some specialists [medical geneticists, paediatricians, neurologists or surgeons (colon cancer)] have organised the follow-up of their own patients [5]. Recurrent courses for rehabilitation and adjustment of families with children with different genetic diseases are organised by the Mannerheim League for Child Welfare and sometimes also by some municipalities.

Dimension 2: Access

Genetic services are a part of the public health service, thus the costs for patients are very low. The service covers genetic counselling, including long-term family analyses,

laboratory investigations, karyotyping, molecular studies and enzyme assays and corresponding prenatal diagnosis.

Local public health care centres, regional hospitals, central hospitals and private practitioners refer patients to the university units or to the department of medical genetics at FFF. People can also contact genetic clinics themselves, which gives more flexibility to the system.

The autonomy in health care priorities by the municipalities leads to uneven availability of genetic services. This mainly affects prenatal diagnosis, i.e. maternal serum screening and the age limits for Down syndrome screening. Private prenatal diagnosis is available in all parts of Finland. Those who do not fulfil the medical indications for public prenatal diagnosis in their own municipality can have chorion villus biopsy (CVS) or amniocentesis for chromosome analysis at their own cost, part of which is refunded from national health insurance.

There is no rate schedule for genetic services, neither for counselling nor for laboratory tests. Each unit decides their prices according to their actual budget and some fluctuation occurs every year.

The nationwide public health care centres, especially the maternity and child care units, cover 99% of the population. Special, unclear and genetic cases are referred to central hospitals which refer to the medical genetics units. This leads to a high rate of identification of individuals eligible for genetic services. Once identified, these individuals have access to all available genetic services if they so wish.

The public health care system is designed to be accessible to people independently of their educational and economic status. Therefore, genetic services that are part of the public system are evenly used by the whole population, including foreigners. On the other hand, more educated people are more likely to use genetic services available upon request, like those at the Department of Medical Genetics at FFF [6].

In rural Finland, genetic satellite clinics are organised by three university hospitals (Oulu, Kuopio and Tampere) in more remote hospitals. The visiting medical geneticist at the satellite clinics provides clinical diagnosis and counselling and organises laboratory tests and prenatal diagnosis to be performed at the university hospitals.

The need for genetic counselling to be properly recognised in primary care has not been studied. Because patients can contact the genetics clinics directly without referrals, the opportunities for everyone to have access to services are very good.

Dimension 3: Life Sustaining

The fertility rate (births per 1,000 women aged 15–49 years) is 51.2. Every year about 65,000 (65,480 in 1994) children are born and about 10,000 (10,031 in 1994) pregnancies are terminated; 14.4% of the mothers are 35 years or older at delivery. The infant mortality (under 1 year of age) is 4.6 per thousand live births (1994). The figure is very low due to effective prenatal diagnosis of severe congenital malformations by ultrasound and termination of these pregnancies. Nevertheless, congenital malformations count for 40% of the infant mortality [7]. Life expectancy is 71.4 years for men and 79.3 for women.

The epidemiology of hereditary non-polyposis colorectal cancer throughout Finland has been published [8]. The cancer register (Stakes) [9] is an official register that contains data on all cancer cases. Many scientists have collected data concerning different genetic diseases, for instance at the Department of Medical Genetics at FFF.

Dimension 4: State of the Art

The speciality of medical genetics was recognised in 1981 and the specialisation programme is run by three medical faculties (Helsinki, Turku and Oulu). In 1982, six medical geneticists gained this competence. Now (in 1996) there are 21 specialists in medical genetics and 10 physicians enrolled in the training programme which lasts 6 years. The Universities of Helsinki and Turku have departments and chairs of medical genetics. Preclinical genetics studies for medical students comprises lectures, laboratory demonstrations and clinical teaching. The medical faculties without departments of medical genetics and the clinical departments have integrated teaching of genetics in their own teaching programmes. The heads of tertiary centres are medical geneticists responsible for the diagnosis of genetic diseases, consultations from other departments and genetic counselling.

Natural scientists (biologists) can specialise in medical genetics in 4 years provided that they have an MSc degree in which genetics is the main topic and which contains a certain amount of human genetics. They are called 'hospital geneticists'. The specialisation programme was introduced in 1990 and six geneticists gained competence during that year. Now (1996) there are 9 competent hospital geneticists, and 20 trainees are approved for the programme and are still in training. The programme is run by the University of Helsinki and approved by the National Board of Medico-Legal Affairs at the Ministry of Social

Affairs and Health. There is a special board that grants competence for those who apply for it after finishing the programme. Laboratories can be run by these 'hospital geneticists' and they usually sign test reports.

There is no organised special training for genetic nurses, who obtain in-service training while working at the medical genetics units. Genetic nurses perform preliminary interviews with patients, collect family data and samples and organise the follow-up for families. In most departments, counselling prior to amniocentesis or CVS, resulting from advanced maternal age or elevated risk for Down syndrome in maternal serum screening, is performed by genetic nurses.

Medical genetics as a speciality in medicine is reserved for physicians. There is no 'certified degree' for medical specialists of other disciplines to acquire and apply medical genetics to their own field. However, it is possible to gain specialist competence in more than one field of medicine.

Training centres must be approved by the boards set up by the universities to run the specialist training programmes. The same boards also approve the genetic trainees, on request. Clinical and molecular genetics and cytogenetics are linked in tertiary centres at four university central hospitals. The fifth has a counselling clinic only but it works tightly connected with the medical genetics departments of the corresponding university (Turku). There is no appointed reference centre.

At the department of medical genetics at FFF and at the Folkhälsan department of medical genetics, genetic counselling is performed outside the hospital system but in close collaboration with the multi-disciplinary teams at the university hospitals. Similarly, five laboratories, including three private ones, work separately. Laboratory equipment has been appropriate, funded either by the public sector or by private funds. Lack of equipment has not been a limiting factor in developing genetic services.

The two national societies, The Finnish Society for Medical Genetics and Geneticists in Health Care (see Dimension 5), arrange meetings and courses for scientific and practical training at least twice a year. Informal meetings for patient reviews, discussions on current clinical practice, laboratory problems and standards are also organised once or twice a year.

Dimension 5: Non-Harmful

Laws Regulating Safety for Patients and Professionals

The fact that medical genetics was recognised as a medical speciality of its own in 1981 gave it an official status which is well known and accepted. The rights and responsibilities of all professionals in health care are regulated by a statute law and decree [10, 11]. According to the Patient Injury Act [12] every employer or private practitioner in health care has to have insurance to cover any complications. The patient can report the injury to the Patient Insurance Association. There, experts are used to judge if there has been an injury justifying compensation, but guilt is not sought. If it is found that the insurance fee has not been paid, it can be collected in six instalments.

Confidentiality and Genetic Registers

Recent developments in confidentiality law [13] have made access to patient medical histories more difficult, hindering genetic counselling particularly if information about family members is needed. In accordance with the confidentiality laws, there are no official genetic family registers. Scientists interested in a particular disease may have their own registers [14] on families included in their studies.

All genetics departments have their own patient registers protected from outsiders. The National Research and Development Centre for Welfare and Health (Stakes) keeps some national registers [15]. The Finnish Register of Congenital Malformations has operated on a population basis since 1963.

Patient-specific information on in-patient care has been collected for the register using a separate report form since 1967. Since 1995, the care report system also covers the institutional care of the elderly and the mentally handicapped, and out-patient care. A medical birth register was started in 1987. There are also registers for causes of death, induced abortion and sterilisation (with indications) and visual impairment. The cancer register is run by the Cancer Society of Finland. If some specific data are to be collected from these registers, approval has to be given by the Ministry of Social Affairs and Health.

Quality

There is no quality control of counselling, carrier screening or prenatal diagnosis but recommendations have been published and updated several times by the Society for Medical Genetics. The Ministry of Social Affairs and Health may recommend policies in medical genetics as in health care in general. But as mentioned, the

communes are free to decide about their policies. Quality assessment has so far been started for cytogenetic laboratories only. The quality control is performed by a private company, Labquality Ltd, which is owned by the largest hospitals and laboratories in Finland. Ten laboratories participated in the 1994 survey and 11 in 1995, including laboratories from Norway and Lithuania. Quality control is conducted biannually.

National Associations

The Finnish Society for Medical Genetics was founded in 1976. Today its main purpose is to organize scientific meetings. It has about 200 members and is open to everybody interested in the field. In 1983, natural scientists founded Geneticists in Health Care, a small association, the main interest of which was to establish the training programme for hospital geneticists and make it a recognised (non-medical) speciality.

Dimension 6: Effectiveness

There are no organised evaluations of the effectiveness of genetic services but there are individual reports of the uptake of prenatal diagnostic tests and the influence on births of children with Down syndrome [6, 16, 17].

Prenatal diagnosis and selective abortion are well accepted in Finnish society [16]. Termination of pregnancy is allowed until the end of week 20 if there is a risk or suspicion of fetal anomaly. If the anomaly is confirmed by reliable prenatal diagnostic means, termination is allowed until the end of week 24. The National Board of Medico-Legal Affairs (TEO) grants permission for these induced abortions (about 220 each year). There are no strong anti-abortion movements in Finland influencing general opinion on prenatal diagnosis. Only some people, mainly religious minority groups are strictly against abortion.

Uptake of prenatal chromosome studies because of advanced maternal age varies from 50 to 85%. The numbers are higher in southern and urban parts of the country. After maternal serum screening, in cases with elevated risk for Down syndrome, the uptake is very high, 98% [17]. Over 95% of fetuses found to have trisomy 21 are terminated. The situation is probably the same for neural tube defects and CNF.

An evaluation of genetic counselling was made by Somer et al. [6]. They studied recall of information, post-counselling reproduction, and the attitude of counselled individuals in families which had received genetic counselling between 1972 and 1981: 62% of the counselled felt that counselling had great or moderate impact on their reproductive plans. Attitudes towards genetic testing of the Finnish population were found to be positive in a nationwide questionnaire study [3, 18]. The value of follow-up for over 10 years in hereditary colorectal carcinoma has been studied [8, 19, 20].

Dimension 7: Consumer Satisfaction

Women's satisfaction with prenatal diagnosis was included in a study concerning maternal serum screening for Down syndrome [17] and most were generally in favour of these studies; 85% of pregnant mothers participated in screening and 95% of these considered the screening test very or quite useful, but more information at all stages of the procedure was requested.

Patient Organisations

There are a large number of lay organisations for various groups of diseases. Some operate under large cover organisations such as the people with short stature under the association for the handicapped, Ehlers-Danlos and some other connective tissue disease groups under the association for rheumatoid diseases, the parents of children with Spielmeyer-Sjögren syndrome (juvenile neuronal ceroid lipofuscinosis) under the central organisation for the visually handicapped, parents of children with cystic fibrosis under the association for lung diseases and groups for some genetic neurologic diseases under the multiple sclerosis association. There is an organisation for parents of mentally handicapped children and another for all people with hearing defects. On the other hand, there are separate associations for families with certain diseases, such as AGU, INCL and retinitis pigmentosa.

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'On behalf of the Finnish Society for Medical Genetics I hereby approve the chapter of 'Assessment of Quality of Genetic Service, Finland' by Harriet von Koskull and Riitta Salonen.'