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Genetic Services in Slovenia

Country Background: Demography, Geography and Infrastructure

Slovenia is a young central European country with a parliamentary democracy which was established in 1991. On December 31, 1995 it had a population of 1,983,012 at 125 people/km^2. The population is relatively homogenous and 87.84% are Slovenes while the most numerous foreign nationalities are the Croats (2.76%), Serbs (2.44%), Muslims (1.36%) and Hungarians (0.43%). The majority of the population are Roman Catholic (84%). The size of families is generally small; 37% of families have one child, 34% two children, 6% three children and 23% more than three children.

Health Service Setting

Expenditure on health care was 7.8% of the GDP in 1995. Individuals pay obligatory National Insurance contributions but they can choose private insurance schemes for non-standard facilities. The health insurance system covers 100% of the population. Planning and supervision of health services is the responsibility of the Ministry of Health. Over many years Slovenia has developed a widespread net of health services at primary, secondary and tertiary levels which is available to all the population. There are 57 hospital beds and 11.5 physicians per 10,000 people. In recent years there has been a tendency to put more emphasis on primary care at the expense of the secondary and tertiary levels. Most medical care is provided by government employees although private practice was legalized in 1993 and based on contracts and accreditations with the National Insurance Institution, 8.12% of GPs and paediatricians and 11.3% of other specialists were in private practice included in the national health service in 1995.

History of Medical Genetics

Karyotyping started in 1958 at the National Institute of Public Health and in 1966 at the Division of Medical Genetics (DMG), Department of Obstetrics and Gynaecology (DOG), University Medical Centre Ljubljana (UMCL). While the first institution remained mainly laboratory based and specialised in mutagenesis analysis, the DMG developed into a tertiary genetic centre providing both genetic counselling and laboratory investigations. Prenatal diagnosis started in the DMG in 1981. Molecular genetic analysis started on a research basis in 1987 at the Institute of Biochemistry, Medical Faculty Ljubljana, and in 1993 in the clinical setting at the DMG, UMCL. Genetic counselling has been provided since 1972 at the DMG, and for some time also at the Department of Paediatrics, UMCL and Department of Orthopaedics, UMCL. Currently, the DMG is the only centre providing genetic counselling in Slovenia.

Dimension 1: Availability

Distribution

Genetic services are provided by one medical genetic centre, three cytogenetic laboratories and four molecular genetic laboratories, for a population of about 2,000,000. Recently, a private institution, which includes genetic counselling and a cytogenetic laboratory for prenatal and

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postnatal diagnosis, was established, and will start operating in November 1996 (see Annex). The DMG is the only institution in the country providing genetic counselling and cytogenetic prenatal diagnosis services. Additionally there are three cytogenetic and four molecular genetic laboratories, mainly specialised in certain aspects of medical genetics. The Annex provides the number of genetic counselling sessions and diagnostic samples analysed in 1995, or in 1996 until November 1, for those laboratories that started operating during 1996. Most genetic testing is interpreted and genetic counselling given at the DMG, while some genetic counselling is still provided by paediatricians (haemophilia) and gynaecologists (cytogenetic testing) who have not received genetic training. The molecular genetics facilities in Slovenia cover the diagnosis of Duchenne and Becker muscular dystrophy, myotonic dystrophy, fragile X, Huntington disease, cystic fibrosis (CF), haemophilia A, polycystic kidney disease, incontinentia pigmenti, T and B lymphomas, whereas prenatal or postnatal diagnosis of other conditions is arranged in other European centres by the DMG.

Neonatal genetic screening is available for the whole population for PKU and hypothyroidism, whereas neonatal screening for CF and the triple hormone maternal serum screening test for Down syndrome have been made available in the period 1995–1996 through research projects. The latter two tests are being proposed to the National Insurance Institution for financing in 1997. Prenatal diagnosis (amniocentesis) has been offered to every pregnant women aged 35 or over since the beginning of 1996. Before then the age limit was 37 years.

Recessive disorders, apart from PKU and CF, are not a major problem in Slovenia, as the level of consanguinity is low, thus no population screening is organised. Prenatal and postnatal biochemical genetic analysis is mainly organised with the collaboration of centres abroad.

Co-Ordination and Integration of Primary, Secondary and Tertiary Provision

The vast majority of patients are referred for genetic counselling by GPs or specialists at the primary care level. The co-ordination of the primary and secondary levels is informal and is based on direct contacts with physicians. At the tertiary level, geneticists and the appropriate specialists hold joint clinics for neurogenetics, while similar clinics are being organised for cancer families and genodermatoses. Helped by the relatively small size of Slovenia, both in population and geography, the overall coordination is relatively efficient.

Long-Term Care Facilities

Patients and their families can obtain long-term care and regular follow-up (referral through GPs or specialists at the primary care level). 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 DMG. The clinical long-term management of patients is provided either in specialised governmental institutions or, in special cases, assisted nurse or apparatus care is provided at the patients' homes.

Dimension 2: Access

The funding of genetic services is included in the general health system. Genetic counselling and laboratory tests suggested by the medical geneticist are free of charge and economic factors do not restrict equity of access to genetic services in Slovenia. However, consumers' access to genetic services is restricted by the relatively poor public awareness of genetic services as well as the sometimes unsatisfactory genetic education of practitioners at the primary and secondary levels.

Patients are referred to genetic services either by GPs or specialists at the primary, secondary or tertiary level. There is relatively good coordination between DMG/DOG and other tertiary medical institutions in Slovenia. The evaluation of patients with rare/genetic diseases is largely centralised in the departments of UMCL.

Dimension 3: Life Sustaining

The number of births is slowly decreasing and total births were 19,751 in 1994. Mean maternal age has been slowly increasing over the past 13 years, and was 26.7 years in 1993. The maternal fertility rate for the same year was 1.31. The infant mortality rate decreased from 14.1 deaths under 1 year per 1,000 live births in 1980–1984 to 6.8 in 1993. Life expectancy was 69.4 years for males and 77.29 years for females in 1993.

Predictive genetic tests for life-threatening disorders are not routinely in use although research on genetic factors in atherosclerosis, risk factors for coronary heart disease and cerebral vascular insult as well as genetic analysis of breast cancer are currently underway. A cancer family clinic is being developed and the Cancer Registry of Slovenia has maintained a database of cancer patients since 1950. The leading causes of deaths in 1993 were circulato-

ry diseases (45%), neoplasms (23%) and accidents, poisonings and violence (9.5%). Congenital anomalies contributed 0.005%. Mortality rates have not changed significantly over the past 13 years (10.3 in 1880 and 10.0 in 1993).

Dimension 4: State of the Art

Medical genetics is not recognized as a separate clinical specialty. The Slovene Association of Human Genetics (SAHG) was founded in 1984 and currently has 142 members. SAHG recently developed a training programme for clinical geneticists (2 years) as well as for medical cytogeneticists and medical molecular geneticists (each lasting 1 year).

Since 1972, human genetics has been taught at the Institute of Cell Biology, Medical Faculty Ljubljana with participation of the DMG. Human genetics is taught at the Medical Faculty Ljubljana in the 2nd year in a 15-hour course. Medical genetics is also included in the curriculum in later years of undergraduate medical studies, incorporated in lectures on gynaecology and obstetrics, paediatrics and neurology.

SAHG has recommended to the National Board of Health the establishment of a training programme for clinical geneticists, medical cytogeneticists and medical molecular geneticists. The course for medical geneticists would last 2 years and entry requirements would be specialisation in a branch of clinical medicine, e.g. paediatrics, neurology, internal medicine. The curriculum would include courses in basic human genetics, medical cytogenetics, medical molecular genetics and syndromology with additional practical training at a clinical genetic centre, and cytogenetic and molecular genetic laboratories. It is desirable that a trainee undertakes research resulting in publication. With respect to genetic counselling, the programme includes genealogical analysis, clinical method in genetic counselling, psychological method in genetic counselling, risk estimation in mendelian inherited/multifactorial/chromosomal disorders and legislation in the field of medical genetics. Trainees should become familiarised with the main topics of clinical genetics in paediatrics, neurology, gynaecology and obstetrics, ophthalmology, dermatology and oncology.

The cytogenetics and molecular genetics training programmes will last 1 year and a diploma in biology, chemistry, pharmacology, medicine or veterinary medicine is required. Programmes are technically oriented with the emphasis on quality control and genetic counselling implications.

Being a small country with one genetic centre, collaboration with physicians at primary, secondary and tertiary levels is good. SAHG is preparing teaching courses for some specialties (general medicine, paediatrics, obstetrics and gynaecology and neurology) to improve the quality of genetic service.

Up-to-date genetic laboratory equipment is relatively adequate for cytogenetic and molecular genetic purposes but there is a shortage of advanced biochemical analysis. However, the efficient organisation of laboratory analyses for a small country with a population of 2,000,000 requires good co-operation with other countries. Medical genetics research is essential for developing molecular genetic analysis in clinical practice and projects are financed by the Slovene Ministry of Science and Technology or international institutions such as the EU.

Dimension 5: Non-Harmful

All health care professionals are supervised by the National Board of Health and can be subject to sanctions for poor practices. A proposal is to be presented to parliament for a law on genetic technology which would regulate also genetic analysis and gene therapy in clinical practice. A national scheme for quality assurance for genetic counselling, medical cytogenetics and medical molecular genetics is being developed. Registration of patient data (partially computerised) is regulated by law and all data are strictly confidential.

Dimension 6: Effectiveness

Genetic counselling is not directive and prenatal and postnatal tests in our country are based on two principles: respect for a woman's free will in deciding about her progeny and the parents' right to medical assistance to have healthy children. A recent evaluation of Down syndrome (DS) for the period 1987–1994 has shown that only 17% of DS pregnancies were diagnosed prenatally. The uptake of prenatal diagnosis in mothers 37 years and older was 35% for the whole study period, but 60% in 1994. In 1996, the age limit was lowered to 35 years. The cost of amniocentesis is around 620 ECU, and 505 ECU for chorionic villus samples.

Pilot neonatal screening programmes for CF and maternal serum triple screening will end in 1996 and results are not yet available.

Dimension 7: Consumer Satisfaction

The social impact of genetic services has not yet been evaluated in Slovenia. There are some patient and parent organisations that cover groups of disorders with a genetic component, such as the Association for Neuromuscular Disorders, Mental Retardation and the Association for the Blind. There are joint activities of these associations with the DMG to keep patients and parents informed about new forms of diagnosis and prevention of genetic diseases. The activities of the genetic interest groups and SAHG will hopefully improve the general awareness of the management and prevention of genetic diseases amongst the general population, patients and their families as well as among health professionals.

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Publications Related to Genetic Services

1 Health Statistical Annual, 1995. Zdrav Vars 1996;35(suppl 5):1–447.
2 Statistical Yearbook, Republic of Slovenia 1994, 33rd issue, Ljubljana 1994.
3 Verdenik I, Pajntar M (ed): Perinatologia Slovenica 1994. Ljubljana, Research Unit, Department of Obstetrics and Gynecology, 1995.

Slovene Association for Human Genetics

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'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.'