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Genetic Services in Lithuania

Country Background: Demography, Geography and Infrastructure

The Republic of Lithuania is a small European country located by the south-east coast of the Baltic Sea. It is 62,300 km^2 in size with a population of 3,798,000 and a population density of 58/km^2; 69% of the population live in urban areas. It has recently regained independence from the former Soviet Union (March 11, 1990) and is a parliamentary republic with a 96% literacy rate. The infrastructure is in need of modernisation and is undergoing economic reconstruction. The majority of inhabitants are Roman Catholic but other important religions are Russian Orthodox, Evangelical Lutheran and Judaism. The ethnic composition of the country is as follows: 79.6% are Lithuanians, 9.4% are Russians, 7% are Poles, 1.7% are Belorussians, 1.1% are Ukrainians; there are also small groups of Tatars, Latvians and Germans. Non-Lithuanians are mostly concentrated in the biggest cities. Most of the Poles and Belorussians live in the south-eastern part of the country.

Health Service Setting

There are 49 physicians and 123 hospital beds per 10,000 inhabitants. Individual and public health services are headed by the Ministry of Health and funded from the national budget. From July 1, 1997, the system is to be reorganised into a National Health Insurance System. Two university, 6 university-associated and republican hospitals, 3 specialised scientific centres and institutes and 24 specialised hospitals, as well as other institutions maintaining and servicing health care, function under the supervision of the Ministry of Health. Other medical institutions are subordinate to self-governments. Health care departments or city hospitals are responsible for health care in cities, while in districts this responsibility rests with the central district hospital.

History of Medical Genetics

The first genetic counselling clinic in Lithuania was founded in 1971 at Vilnius Republican Clinical Hospital. In 1986 it was reorganised as the Centre of Medical Genetics and in 1991 it was reorganised as Vilnius University Human Genetics Centre.

Dimension 1: Availability

Table 1 details personnel involved in genetic services in Lithuania. Prenatal diagnosis was started in medical genetic services in Lithuania in 1987. Vilnius University Human Genetic Centre is the only institution in Lithuania where congenital fetal diseases are detected by invasive procedures for fetal chromosomal analysis and single-gene disorders. More than 2,500 pregnant women from the whole country are examined by ultrasound there annually and about 150 chorionic villus samplings (CVS) are performed (144 CVS in 1995). The procedure of amniocentesis is not performed.

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Table 1. Medical genetics staff in genetic service provision in Lithuania

Number of centres	Population served	Genetically trained physicians	Genetic physicians in training	Cyto-geneticists	Molecular and biochemical geneticists	Genetic nurses/ counsellors
1	3,798,000	8	2	2	7+5	4/0

Placentocentesis (second and third trimester placental biopsy) was included in an ongoing first trimester transcervical CVS programme in 1993. Since 1994, all CVS have been performed transabdominally, a change which has been associated with a reduced fetal loss, from 4.6% in the transcervical group to about 0.5% using the transabdominal route. Placentocentesis is used in chromosomal analysis of complicated fetal abnormalities detected by ultrasound examination.

Legal abortions are allowed until 12 weeks of gestation. Pregnancy termination for medical indications (fetal non-viability) is legal until 22 weeks of gestation.

A specialised cancer genetics service network has not yet been established.

The Ministry of Health sponsors a national programme for the detection and prevention of birth defects. Nationwide neonatal screening programmes for PKU and congenital hypothyroidism (CH) have been in action since 1975 and 1993, respectively. Neonatal screening for CH was discontinued for 8 months in 1996 due to temporary cessation of funding. PKU treatment is centralised in Vilnius University Human Genetics Centre. In the case of suspected disease, newborns visit this centre at about 21 days of age and stay in the hospital for confirmation of the diagnosis. In addition, molecular screening for fragile X syndrome, nine cystic fibrosis mutations and the triple test (α-fetoprotein, beta-HCG and E3) are available at present.

The main tasks of the Human Genetics Centre in Lithuania are genetic counselling, neonatal mass screening for PKU and CH, prenatal diagnosis of congenital anomalies, and the Lithuanian Registry of Inherited Diseases and Congenital Anomalies (LIRECA). The Human Genetics Centre is the only genetics centre in the whole country. No other public or private genetic laboratories exist.

All patients referred by a general practitioner or other medical professional are counselled by a genetically trained physician (clinical geneticist), to be examined by trained medical geneticists in cases of the aforementioned diseases (staff of the Laboratory of Molecular Genetics) or in case of a suspected chromosomal disease (staff of the Laboratory of Cytogenetics). Prenatal biochemical screening is performed in cases of pregnancy at risk. Prenatal screening for advanced maternal age is performed from 40 years of age. Newborn screening for PKU and CH is mass screening. Population screening for carriers of recessive disorders is not performed. The genetic register, all laboratories and the genetic counselling clinic are at the Human Genetics Centre in Vilnius.

Dimension 2: Access

Restrictions to access are as follows: (1) poor funding for the health service in general; (2) most practitioners trained before 1991 have a poor knowledge of genetic diseases; (3) the patient who sees a geneticist without first being referred by a general practitioner or other medical professional must pay for the counselling and the tests; (4) at present, a significant proportion of the Lithuanian population have a very low income and so are reluctant to spend money on travel expenses, and (5) much of society considers genetic diseases to be incurable.

Dimension 3: Life Sustaining

The birth rate per 1,000 population in 1995 was 11.5, i.e. the same as in the early 1960s. The average life expectancy has been increasing slightly in the last 20 years but by the beginning of 1991 the process had been reversed. Female life expectancy differs little from that in highly developed countries (74.93 years), whereas the average male life expectancy is 10 years shorter (62.79 years).

Decreasing numbers of births and increasing numbers of deaths lead to negative natural increase. Due to the decreasing birth rate and a comparatively long life expectancy, the population of Lithuania is ageing, thus creating social and health care problems.

The infant mortality rate in 1994 was 13.8 per 1,000. The index of infant mortality has been steadily decreasing

during the last 20 years. In 1991, newborn babies started to be registered according to the recommendations of the World Health Organisation.

At present there is little information on inherited diseases in Lithuania although several genetic investigations have been carried out. Attention was concentrated mostly on chromosomal diseases, genetics of allergic and other diseases, congenital anomalies and inherited metabolic diseases. The total number of pregnancies in 1995 was 41,624, with 447 (1.07%) where the mother was aged 40 or over.

Dimension 4: State of the Art

The Lithuanian Society of Human Genetics was established in 1991. At the moment there are 28 members who are personally engaged in the field of human and medical genetics and 40 specialists. Not all specialists employed in the Human Genetics Centre at Vilnius are members of the society. Those who are not are either clinical geneticists or medical geneticists with a university education. Besides those mentioned in table 1 there are also mathematicians, computer specialists, economists, psychologists, laboratory assistants and nurses without genetic qualifications. The society organises training and teaching in human genetics at home and abroad and assists in the promotion and publication of scientific work of its members. One of its most well-known activities is organising courses in human genetics. One-week intensive courses in human genetics were organised in Lithuania in 1984, 1989, and 1994. Each participant was obliged to deliver a lecture.

A number of scientists from western countries support medical genetics in Lithuania. They carry out some free-of-charge analyses for prenatal diagnosis in Lithuanian families, accept Lithuanian specialists for training courses in clinical and molecular genetics, assist in purchasing necessary equipment and reagents for the research carried out under CA EU programmes and provide (most often as a personal gift) the Human Genetics Centre in Vilnius with medical and scientific literature including subscriptions to some scientific journals.

At present, only one school, namely the Medical Faculty of Vilnius University, provides training for specialists in human and medical genetics. This centre is also responsible for training Vilnius University medical students, residents, postgraduate and PhD students, as well as scientific research. The main research interest is human genome diversity. In recent years, molecular genetic investigation of inherited diseases, such as PKU, cystic fibrosis, retinitis pigmentosa, Duchenne muscular dystrophy, fragile X syndrome, has been in progress in the Human Genetics Centre. Unfortunately, financial problems place severe constraints on such investigations. Molecular genetic investigation of familial adenomatous polyposis was introduced in Lithuania at the beginning of 1995.

The courses in human genetics are for specialists with a university education, i.e. for clinical geneticists (university education, MD or PhD in medicine), medical geneticists (university education, MSc or PhD in genetics or biochemistry) and other physicians or biologists interested in the problems of human and medical genetics. These courses are affiliated to Vilnius University and participants receive a certificate.

Since 1990, genetic teaching to medical students has included the following course: introduction to genetics to 1st year medical students (16 h), human genetics (16 h theory, 32 h teaching group practice) in the 8th semester and clinical genetics for 6th-year students (32 h teaching group practice). This last course includes lectures on genetic assessment and counselling, classification of congenital and hereditary diseases, aspects of teratology, congenital eye diseases, genetics of mental retardation, congenital endocrine system disorders and a model approach to cystic fibrosis, as well as group teaching on the most important points of chromosomal and single-gene disorders, congenital malformations, prenatal diagnosis, the LIRECA and ethics in genetics.

Since April 29, 1991, clinical genetics has been a medical specialty recognised by the Ministry of Health and special training programmes have been introduced. Six years of undergraduate training are followed by 1 year of compulsory residency. Special training in clinical genetics takes another 2 years. Until 1991, clinical geneticists, after 6 years of undergraduate university training and 1 year of an internship, usually went to Moscow for a 3-month course in clinical genetics. Now they receive special training in clinical genetics within the country which takes 2 years for residents. Although self-education is continuous, updating knowledge on postgraduate courses is essential. There is a lack of information about such courses in other countries, although even when enrolled, Lithuanian specialists are unable to pay for the courses, tuition and travel, and their home institutions are unable to support them financially. The help of western countries in professional training is ad hoc and depends on personal contacts.

Nurses and midwives are trained in five medical schools in Lithuania and receive training in elements of

human and medical genetics. Specialised genetic nurses and genetic counsellors are not formally trained but before starting their activities as genetic nurses in the Human Genetics Center, nurses have a 3-month course of lectures and group teaching on the most important aspects of clinical genetics and defined techniques.

Dimension 5: Non-Harmful

The LIRECA, with total population coverage, has been in operation since 1992. It is a hospital-based surveillance system of congenital anomalies occurring in newborns, stillborns and abortions. Completion of notification forms is compulsory and case records are updated when necessary from multiple sources of information. Diagnoses are made by neonatologists, paediatricians, geneticists, cardiologists and pathologists. Notification forms are filled in in all newborn departments, delivery units, pathology centres, child cardiology centres, specialised newborn centres and the Human Genetics Centre. The data on congenital anomalies and patients is computerised and kept confidential in the Human Genetics Centre, which is the only institution in the Republic of Lithuania engaged in the field of medical genetics.

The following factors reduce the quality of genetic services in Lithuania: before 1991 general practitioners were untrained in genetics; clinical geneticists have limited opportunities for updating their knowledge in human and medical genetics; there are limited possibilities for clinical geneticists to offer up-to-date genetic tests for patients (due to poor funding in general).

Dimensions 6 and 7: Effectiveness and Consumer Satisfaction

No information was available on these two dimensions for Lithuania.

Publications Related to Genetic Services

No information available.

Lithuanian Society of Human Genetics

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'I am writing to confirm that the data presented in the manuscript of V. Kučinskas 'Genetic Services in Lithuania' are valid.'