European Journal of Human Genetics

Eur J Hum Genet 1997;5(suppl 2):178–182

E. Tunçbilek

Division of Clinical Genetics, Hacettepe University, Faculty of Medicine, Children's Hospital, Ankara, Turkey

Genetic Services in Turkey

Country Background: Demography, Geography and Infrastructure

Turkey has a surface area of 774,815 km^2 and a land area in both Europe and Asia. About 3% of the total area lies in south-eastern Europe (Thrace) and the remainder in south-western Asia. The latest census in 1990 put the population of Turkey at 56.5 million and the population growth rate was 21.7 per 1,000 for the 1985–1990 period. The proportion of urban population was 56%. The total population estimate for 1995 is 61.4 million. Turkey has a young population structure, a third of the population is under 15 years of age, while the proportion of the elderly is quite low. Marriage, predominantly civil, is widely practised in Turkey. Consanguineous marriages also account for a significant proportion of the marriages at 22.6%; 66.3% of these consanguineous marriages are first cousin marriages. Turkey has a parliamentary, multiparty system and the literacy rate is 81%. There are diverse geographical, climatic, cultural, social and economic differences in the country which to some extent reflect the differences in socio-economic development levels and demographic conditions among regions of the country. Citizens of Turkey are predominantly Muslim (98%). Ethnically, Turks predominate; Kurdish, Arabic, Greek, Circassian, Georgian, Armenian and Jewish communities of varying sizes complete the ethnic mosaic of the rich and complex culture of Turkish society. This diversity of Turkey's population is also very challenging for genetic studies.

Health Service Setting

Health care in Turkey is provided by a number of separate agencies. Two organisations are involved in formulating health policy: the Ministry of Health (MoH) and the State Planning Organisation (SPO). Other important organisations such as the Social Insurance Organisation (SSK) and the university hospitals pursue particular objectives but do not in fact determine policy.

Health services provided by the MoH are administered by Provincial Health Directorates. Below this level there are two types of health organisation. The first, which started in 1963 in the eastern provinces and covered the whole country by 1984, accounts for a programme of integrated and curative services provided without charge by health centres. As satellites to these health centres, midwife stations or 'health houses' for maternal and child health care have been established. Besides these integrated services, some vertical health organisations still exist to run health programmes for e.g. tuberculosis, mother and child care, and family planning.

Beyond the provision of primary health care, the MoH scheme maintains a large network of hospitals. However, these hospitals contain only half of the national total of beds. In Turkey, there are 9 doctors and 22 hospital beds per 10,000 population. Currently, the government is working on a system in which family physicians will work by contract with the health insurance organisation. The hospitals will be decentralised and privatised. This process will start with the state-owned hospitals.

In Turkey, a large number of agencies are involved both as providers and financial intermediaries. The private sector accounts for approximately 55% of total expenditure, mostly for non-hospital services. MoH institutions are responsible for about 20% of total expenditure. On the funding side there are three main sources: the general state budget (±30%), the health insurance funds (20%) and out-of-pocket payments by users (50%).

Among the social security programmes, the SSK is the most important. SSK originally covered industrial and commercial workers (blue collar workers). The SSK has a

KARGER

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

E. Tunçbilek
Division of Clinical Genetics
Hacettepe University
Faculty of Medicine, Children's Hospital
TR–06100 Sihhiye, Ankara (Turkey)

Table 1. Staff engaged in genetic service provision in Turkey

Number of centres	Population served	Genetically trained physicians	Genetic physicians in training	Cyto-geneticists	Molecular geneticists	Genetic nurses/ counsellors
26	61.4 million	14	20–30	49	28	0

large network of its own service facilities, i.e. hospitals and clinics. The second largest programme is the retirement fund, which covers government employees (white collar workers). The third is social security for self-employed persons and employees such as merchants, artists and farmers. Turkish civil servants at both central and local levels are covered by a separate social security programme.

Altogether, about 50% of the entire population is covered by the social security protection for health services. Those who cannot benefit from social security protection can rely on services from MoH facilities. There are small hospitals that belong to different religious groups in Istanbul. Genetic screening services, such as PKU and hypothyroidism, are free of charge.

History of Medical Genetics

The first genetic units were established in 1964 at Hacettepe Medical Faculty, Ankara and Cerrahpaşa Medical Faculty, Istanbul. All genetic units were located in departments of paediatrics and their interest was mainly clinical genetics and cytogenetics. After the 1980s, departments of biology became involved with molecular genetics.

Although the first laboratory and unit dealing with genetic problems was established in the early 1960s, progress in genetics was rather slow in Turkey. During the 1990s, with the development of molecular methods for treatment and diagnosis of various diseases all around the world, increased concern in molecular genetics emerged in Turkey. Several people have been abroad for education in genetics and returned to serve in Turkey. This concern enhanced the establishment of new services that will provide new techniques for various diseases in Turkey.

Dimension 1: Availability

Genetic departments in Turkey do not serve predetermined districts and any patient from any part of the country is accepted even without referral. This is the main reason for lack of patients registration.

Nearly all genetic services are located in the universities with 22 departments of genetics in Turkey (table 1). A genetics laboratory based in Ankara was established in 1994 in one of the largest maternity hospitals. Three private laboratories in Ankara and Istanbul for cytogenetic tests and one in Istanbul for DNA tests were established in 1995.

Although medical genetics has been an officially recognised speciality since late 1990, the infrastructure of departments giving a qualification in medical genetics is insufficient both in respect to number of academic personnel and equipment, and results are not satisfactory. By 1996, only 14 medical doctors had this qualification. Eighty-one individuals have PhDs both in medical biology and genetics and there are 104 with masters degrees.

The number of medical specialists working in genetic services, but not necessarily certified, is around 18 and the number of scientific personnel working in the field of medical genetics is around 50. There is no officially certified programme in cytogenetics or molecular genetics. Most individuals working in these areas are 'self-declared' and the exact number cannot be stated. Collaborative work sponsored by the Turkish Association of Medical Genetics is in progress to provide exact numbers.

Laboratory facilities for genetic services vary greatly but the following are available in specialised laboratories: cell culture facilities, PCR, oligosynthesisers, personal computers linked to Internet. There are three major molecular genetic laboratories dealing with DNA diagnostics. Hacettepe Universities in Ankara and Bogazici University and DETAM in Istanbul handle more than 500 samples per year. Other main interests are concentrated on haematological disorders such as thalassaemia and sickle cell anaemia, Duchenne muscular dystrophy

(DMD), cystic fibrosis, PKU and spinal muscular atrophy. An additional centre at Bilkent University, Ankara, was established in 1995 for cancer genetics. The high frequency of PKU and thalassaemia has driven the establishment of diagnostic molecular genetic units with clinicians in associated fields. However, there is not yet a network system for co-ordination of the investigation of rare disorders.

In 25 laboratories, routine cytogenetic analysis of peripheral blood can be carried out using various banding techniques. The number of analyses done in a year varies between 0–250 in 16, 250–500 in 6 and more than 500 in 3 laboratories (Hacettepe in Ankara, PRETAM in Istanbul and GENTAM in Eskisehir). Cytogenetic analysis is performed in various universities for various haematological disorders, including Fanconi aplastic anaemia. Turkey is one of the clinical co-ordinators of non-EU countries for the European Fanconi Anaemia Research Project (EUFAR). Cytogenetic analysis of various leukaemias is performed in most cytogenetic laboratories.

There is a concentration of laboratories in western and central Anatolia and there are no major regional laboratories in eastern and south-eastern Turkey. At the moment, patients in these areas are served mostly on a referral basis to various fairly distant laboratories. Although there is no specific laboratory designated for biochemical genetics, various enzyme analyses, especially for neurodegenerative diseases, are performed at Hacettepe and Istanbul Universities.

Various screening programmes have been launched in the last 15 years. A nationwide neonatal PKU screening programme was started in Hacettepe University in 1983 and it continues with the collaboration of several university and government hospitals and clinics under the organisation of the MoH. Hypothyroidism screening was added to this programme in 1991. Screening for various haemoglobinopathies is performed in primary and high school children. Premarital screening was started in 1990 in a city located in a geographical area with a high incidence of HbS. Thalassaemia screening in pregnancies of couples at high risk is being carried out in Hacettepe, Ankara; Çukurova, Adana and Bogazici, Istanbul. Screening for Down syndrome for high-risk pregnancies is available on request by using maternal serum α-fetoprotein and fetal karyotype analysis mainly in six centres (Hacettepe, Gulhane Military Medical Academy and Gazi in Ankara, PRETAM in Istanbul, GENTAM in Eskisehir, Ege in Izmir). At least six other clinics offer prenatal diagnosis for Down syndrome but serve less than 100 patients a year. Screening for neural tube defect by maternal serum α-protein and ultrasonography during pregnancy is common practice in Turkey.

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

Because most medical genetic units are located in university hospital clinics, there are many joint clinics with, for example, clinics for DMD patients with the department of neurology, thalassaemia with the department of haematology, and counselling about the consequences of consanguinity with the department of public health. Training of primary health care personnel and reaching the public through the media are also relevant.

Long-Term Care Facilities

There is no organised system for long-term follow-up of individuals who are at risk for serious genetic disorders. However, collaboration with the department of physiotherapy for DMD patients and with the department of child development and education for patients with Down syndrome can be mentioned as long term rehabilitation of some genetic disorders.

Dimension 2: Access

Genetic services are funded mainly by the government for physical facilities, salaries, some equipment and instruments. Additional funding is available from various governmental and international agencies on a project basis. Consumers have access to genetic services in the same way as general medical care.

The number of genetic specialists is quite low and they are concentrated mainly at large university centres either in western or central Turkey. Therefore there is a physical problem of access. In addition, there is no established system for general practitioners trained in basic genetics to facilitate the referral process. The public is not very aware of genetic problems and their consequences. Turkey's population is very large compared to most European countries, and this is a source of service shortages.

Dimension 3: Life Sustaining

According to the 1993 Demographic and Health Survey of Turkey, the infant mortality rate is 53 per 1,000 live births and more than half of the infant deaths occur in the first 4 weeks of life. Although the probability of death between birth and the 5th birthday (child mortality rate)

is around 61 per 1,000, the survey shows that mortality risks during infancy and childhood have been declining relatively quickly. The decline in infant mortality is 35% in the last 10 years. Life expectancy, according to the 1990 Census of Population, for males is 67 and for females 71.

Dimension 4: State of the Art

There are three types of educational programme in genetics in Turkey.

Speciality Training in 'Medical Genetics'

The applicants are graduates of medical schools and should pass a nationwide entrance examination set by the Universities Selection and Placement Centre (a government organisation). The duration is 2 years but there is no system to standardise the curriculum.

PhD Programmes in Medical Genetics

These are available in four universities (Istanbul, Hacettepe, Ege and Ankara) provided by the departments of paediatrics of these universities under the Institute of Child Health. At Ankara University, a PhD in medical genetics is provided by the Department of Medical Biology. Only medical doctors are accepted in these programmes.

PhD Programmes in Medical Biology

These programmes are offered by 23 medical biology departments which operate under the Institutes of Health Sciences (each Faculty of Medicine has an Institute of Health Science to co-ordinate postgraduate education in basic medical sciences).

There is no standardisation of the contents of PhD programmes. In addition to these PhD programmes, there were two additional programmes leading to a masters degree in medical cytogenetics and genetic counselling at Istanbul University. However, the graduates of these two programmes had difficulty finding employment and these programmes were ended.

At the present time, there is no program for teaching genetics to primary health care staff such as nurses/midwives.

Recently, the Scientific and Technical Research Council of Turkey has indicated its policy to establish genetics, molecular biology, biotechnology and scholarship programmes as priority areas for funding. This may encourage research and the creation of critical mass in these fields.

Dimension 5: Non-Harmful

The MoH is responsible for licensing medical and other health care professionals and laboratories. However, once licensed, neither professionals nor laboratories are subject to routine quality assessment. The High Medical Council working under the MoH judges malpractice in case of complaint but is not responsible for maintenance of quality standards. Societies have been established for research and family care for genetic diseases and certain other disorders, such as PKU, DMD and Down syndrome. There are local review meetings organised by these societies. Guidelines for molecular technology are established within the internal standardisation and quality control systems of each unit. Co-operation with European as well as non-European countries and the USA is organised through joint projects and consortium efforts. Transfer of technology to Turkey is established through such co-operation.

Dimension 6: Effectiveness

Desirable outcomes of medical genetics are those that lead to improvements in the health of the population and can be defined as effective screening and presymptomatic diagnosis of genetic diseases, effective prenatal diagnosis and counselling to create public awareness of genetic problems. Unless the incidence and prevalence of genetic diseases are known it is very difficult to measure desirable outcome. However, by notifying families about the risk of consanguineous marriages and by using all means for public education, the percentage of these marriages decreased to 20% in 1993 from 30% in 1963. Therefore, a decrease in the incidence of common autosomal recessive diseases is expected.

Dimension 7: Consumer Satisfaction

There have been no surveys of consumer satisfaction or of the extent to which consumers have been informed. However, there are frequent programmes on radio and TV, seminars and conferences on patients' rights and ethical issues. No study has been undertaken to assess public acceptance of genetic service but genetics is accepted as part of the medical services. There are no well-established genetic interest groups although societies for various disorders include families and this increases co-operation. Parents' organisations are mostly active in education of mentally handicapped children due to genetic diseases.

Publications Related to Genetic Services

No information available.

Turkish Society for Human Genetics

Prof. Ergül Tunçbilek
Hacettepe University Faculty of Medicine
Department of Paediatrics
Division of Medical Genetics
TR–06100 Hacettepe/Ankara (Turkey)
Tel. 90 312 311 55 22
E-mail etuncbilek@genetic.gen.hun.edu.tr

Validators

Prof. Dr. Meral Özgüç
Hacettepe University Faculty of Medicine
Department of Medical Biology
TR–06100 Sihhiye, Ankara (Turkey)
Tel. 90 312 3110777
Fax 90 312 3110777
E-mail mozguc@genetic.gen.hun.edu.tr

'I find it very beneficial and want to state that it is the only example from Turkey which has collated substantial data on this subject. The author has tabulated the reference data available to her to the best of her knowledge. I would like to extend my validation of the paper.'

Prof. Dr. Isik Bökesoy
Ankara University Faculty of Medicine
Head of the Department of Medical Biology
TR–06100 Ankara (Turkey)
Tel. +90 312 3103010/388
Fax +90 312 3100805
E-mail ibokesoy@dialup.ankara.edu.tr