European Journal of Human Genetics

Eur J Hum Genet 1997;5(suppl 2):163–168

Genetic Services in Spain

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Country Background: Demography and Geography

Spain occupies four-fifths of the Iberian peninsula, the southernmost and the westernmost point of the European continent. Because of its strategic geographical location – a bridge between Europe and Africa – the original native inhabitants, Tartessos and Iberians, were invaded by the Phoenicians, the Celts, the Greeks, the Romans, the Barbarian tribes and the Moors. This circumstance, together with the close relationship with other Mediterranean cultures, determines the unique character of the Iberian peninsula. Spain has 17 regions with their own parliaments that enjoy a considerable degree of autonomy. It has a total area of 505,990 km^2 and 38.9 million inhabitants (77 people/km^2), varying in density from 21 people/km^2 in Castilla-La Mancha to 616 in Madrid. 60% of the population live in areas with more than 10,000 inhabitants, 18% in areas with 2,000–10,000 people and 16% in rural areas [1]. There is not much racial diversity in the population, which is European in origin, with only one distinct ethnic group, the Gypsies, who constitute a small, but important proportion of the population (approximately 1.5%) [2].

Health Service Setting

The organisation of health care in Spain is based on the General Health Law which was passed in 1986, and which regulates the National Health Service (NHS) by integrating different public institutions and agencies. During the last decade, the NHS has been strongly decentralised and health competencies have been transferred to 7 of the 17 autonomous communities: Andalucia, Basque Country, Canarias, Catalonia, Galicia, Navarra, and Valencian Country. Thus, the NHS is formed by the overall health services of the central administration operating through Instituto Nacional de la Salud (INSALUD), and the autonomous communities operating through their own Regional Health Services.

The Spanish health system is currently financed largely by general taxation (80%) and specific social security contributions (20%). Since health financing has been decentralised, half of the health care budget is managed by the autonomous regions. The public sector covers most outpatient health care, and runs over 80% of all hospital beds in the country. The public health insurance system accounts for three-quarters of the total health care expenditure. A private sector exists along with the public scheme and covers about 6% of the population.

Primary Care

The General Health Law of 1986 aimed to implement a system based on a network of health centres in primary care. The geographical and administrative unit in which health services are planned and managed is provided in the primary health care district, with a population varying from 40,000 to 200,000 people. In each district, several health centres will be operative, each staffed with general practitioners, paediatricians, nurses, midwives, social workers and ancillary staff for a population varying from 5,000 to 25,000 people. Nowadays, about 60% of the population is covered by these health centres, varying from 80% in some areas to 20% in others. Therefore, two primary health care systems coexist today. In the old system, general practitioners work from large centres (ambulato-

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rios) or from small satellite centres (consultorios) and combine private practice with very small appointments of 2 h a day in the public system.

History of Medical Genetics

The incorporation of genetic services into medicine in Spain can be dated to the middle of the 1960s, when some health centres began to perform cytogenetic tests, principally aimed at diagnosing congenital defects and haematological disorders. In 1977, the National Plan for the Prevention of Handicap [3] was drawn up under the Royal Patronage of Her Majesty Queen Sofia. The plan had two main goals: (1) to consolidate the existing genetic units/centres, extending their coverage to areas that were not able to meet the demand for genetic services for that population, and (2) to implement a national screening programme for phenylketonuria (PKU) and hypothyroidism. The duration was for one period of 3 years. In 1985, attempts were made to set up a nationwide programme for prenatal diagnosis of chromosomal disorders [4]. Unfortunately, the programme was never fully implemented, although some genetic units were partially financed to extend their coverage of genetic services.

The development of genetic services has been a continuous process but lacked strategic planning. Most units/centres initially emerged as a subspecialty of other medical fields (e.g. biochemistry, obstetrics, paediatrics, clinical pathology) because of the personal interest of individual professionals. Exceptionally, some Regional Health Services have been specially sensitive to the need and potential demand for genetic services in their communities, providing the means to improve the coverage and the quality of such services. The increasing demand during the last decade has greatly contributed to consolidating the existing units/centres and also to the differentiation of services, in many cases from the original medical specialty. At the present time, 65% of the public genetic units/centres are functionally independent, while the rest are structurally organised as part of other departments. This circumstance makes it difficult to estimate the exact number of professionals working in the field.

Responsibility for organising genetic services belongs to INSALUD and the Regional Health Services of the seven communities to which competencies have been transferred. In all regions, however, these services are basically provided by the large hospitals in the public health sector (third level), less frequently by merged private hospitals, and are virtually non-existent at the primary care level. As is the case for other health services, the corresponding costs are mainly covered by the National or Regional Health Services. In the last decade, however, research grants funded by national, regional or local sources, mainly the Fondo de Investigacion Sanitaria, have played an important role in the technological development of genetic services. It can be said that this has contributed greatly to implementing molecular genetic techniques and their application to the diagnosis of genetic diseases.

Dimension 1: Availability

According to information provided by the Spanish Association of Human Genetics, there are currently 37 genetic units/centres performing diagnosis. This does not include private centres that are not associated with INSALUD or the Regional Health Services, nor those basically concerned with research. The personnel of these 37 units/centres include over 150 genetically trained graduates (approximately 50% physicians and 50% non-medical scientists) and 145 technicians or laboratory auxiliaries. One-third of graduates have temporary contracts. About 80 students supported by research grants (17 units/centres) collaborate in diagnostic tasks as part of their genetic training. Practically all units/centres report that they provide genetic counselling services, 83% perform cytogenetic analysis for prenatal diagnosis and over 50% provide molecular diagnostics. Cancer genetics is largely restricted to the diagnosis and prognosis of haematological disorders. The following workloads were reported by 30 of the 37 units/centres during 1994: 15,000 consultations, 19,000 cytogenetic analyses, as well as an undetermined number of molecular analyses for common genetic diseases. With respect to genetic counselling it is important to mention that consultation is often limited to prenatal diagnosis and no more than 10 genetic clinics offer, at the present time, services involving diagnosis, counselling and follow-up of different genetic disorders. However, at least 20 units/centres could actually provide counselling involving a specialist competent in genetics if appropriate facilities and adequate planning were provided by their centres and the National or Regional Health Services.

Predictive diagnosis of inherited disorders, such as Huntington disease, has been incorporated into the diagnostic activities of some genetic units/centres. Medical long-term follow up of patients can also be provided by neurologists, psychologists and clinical geneticists. However, adequate social support and living facilities for peo-

ple at an advanced stage of the disease are not presently available.

Diagnostic services for inborn errors of metabolism are performed in 10 laboratories, which are located in Barcelona (3 centres), Bilbao, Murcia, Madrid, Santiago de Compostela (La Coruña), Sevilla, Valencia and Tenerife. The authors do not have information on the personnel who work in these laboratories, nor the number of samples they analyse. Biochemical analyses, corresponding to the neonatal screening programme for PKU and hypothyroidism, are carried out in 21 laboratories dispersed throughout the country. The communities of Madrid and Murcia also perform screening for 21-hydroxylase deficiency.

It is virtually impossible to determine the provision of prenatal screening for Down syndrome (DS) and neural tube defects (NTD), as they are, in most cases, provided by scattered obstetric clinics that may or may not work in co-ordination with a genetic unit/centre. Currently, two autonomous communities (Basque Country and Catalonia) are evaluating the feasibility of a population-based maternal serum screening programme for DS and NTD, while a smaller region (Navarra) and the area of Móstoles (Madrid) have been offering these services since 1993 and 1991, respectively. It is also worth highlighting the NTD screening programme in Asturias that has been in existence since 1987.

Continued advances in genetic diagnosis for different pathologies have favoured intra- and interhospital relationships between geneticists and other medical specialists, mainly paediatricians, neurologists, haematologists and gynaecologists. These relationships extend, on occasions, to research, planning and the carrying out of joint projects requiring co-ordination and co-operation between professionals at secondary and tertiary care levels. Unfortunately, co-ordination with primary care is practically non-existent. Similarly, official links and co-ordinated genetic programmes with the NHS are very few and limited to biochemical genetics (neonatal screening); co-ordination and planning with the Regional Health Services seems to be more fluid, although no official links are presently established.

There is a good relationship between genetic centres, mainly because of the public social security system that financially covers a large part of the genetic services, allowing them to establish labour relationships without the limitations imposed by specific payment for each analysis.

There are no organised systems for long-term follow-up of patients with genetic disorders. Links between the corresponding medical specialist and the clinical geneticist have not been officially established. Similarly, there is no organised link between specialists and primary care. There is, however, some experience on long-term follow-up of metabolic diseases and, less extensively, for cystic fibrosis and fragile X syndrome in some regions of the country.

Dimension 2: Access

Neonatal screening for PKU and hypothyroidism covers practically 100% of the population, but the quality and availability of other genetic services varies from region to region. Access to genetic units/centres is similar to medical specialities. Patients are referred by general practitioners or specialists to the nearest genetic unit/centre in their autonomous community; if that service is not available, patients may sometimes be referred to a centre in another region of the country. Coverage by genetic services is good in some regions, mainly around large cities. However, there are other areas where the scarcity of genetic centres not only limits access but also impedes education and awareness of the current possibilities for prevention of congenital defects and inherited disorders, for health professionals as well as the general population. Theoretically, there is no discrimination in access to genetic services. However, the information that the consumer has about medical genetics may be very limited and consumers with higher social and education levels have easier access to information on genetic services. Although, since 1986, voluntary interruption of pregnancy has been legal, in some parts of the country, the conscientious objection of a number of health professionals within the public health network in fact limits access to those services.

Financial cover has not been, up to now, a barrier to access to genetic services. Public funding covers the costs of all genetic studies that are medically indicated, including fetal and postnatal karyotyping as well as biochemical and molecular analysis of familial genetic disorders. In practice, however, there are important limiting factors. These include overload of genetic services within the public health sector (there are too few posts, because medical genetics is not recognised as a medical specialty) and inadequate finance for developing molecular genetic services. These not only impede access to genetic services but they also interfere with research, which is essential for the development of human genetics and its clinical applications.

Dimension 3: Life Sustaining

The number of births in Spain is decreasing rapidly [5] with the lowest fertility rate of European countries (1.3 in 1992) [6]. Since 1986, the highest fertility rate has been in mothers aged 30–34 years. In 1992, the mean maternal age was 28.4 years, the average age at first birth was 26.9 years and 11% of mothers were 35 years or older [5]. Partial data obtained from different regions of Spain (national data are not available) show that the proportion of mothers 35 years of age and older is presently higher than 15%. The infant mortality rate has fallen during the last two decades; in 1975, the estimated rate was 18.9 per 1,000, while in 1985, 1990 and 1992 it was 8.9, 7.6 and 7.1 per 1,000 (5.0% during the neonatal period), respectively [6]. Life expectancy is increasing, varying from 70.4 to 73.4 years for men, and from 76.2 to 80.5 for women in 1975 and 1990, respectively [6].

Technological advances in molecular biology have been important in the general development of health care in Spain. During the second half of the 1980s, new DNA techniques for diagnosing monogenic diseases (e.g. cystic fibrosis, myotonic dystrophy) began to be used. At the same time, there was increasing awareness by health professionals about the potential for this technology in diagnosis, prevention and possibly treatment of genetic disorders. Currently, pre- and postnatal and presymptomatic testing and carrier detection for some hereditary disorders are available in some genetic units/centres. Only Huntington disease testing is subject to participation in a programme with a specific protocol. Experience of predictive testing for cancer is very limited. Recently, a few centres have begun providing predictive testing for breast and colon cancers to a limited number of families with documented family history and who in most cases had been previously studied for research purposes.

Dimension 4: State of the Art

One of the main drawbacks for genetic services in Spain is the lack of official recognition of medical genetics as a specialty. This has always been a negative factor in the development of the field, as it makes training of new professionals and their access to employment enormously difficult. The development of genetics in Spain has been largely attributable to the personal interest and motivation of a few medical and non-medical scientists who were able to get financial support mainly through research grants to implement genetic services and new diagnostic technology. Teaching of genetics to medical students was virtually non-existent until 1990. Since then, and according to the 'Real Decreto' (1417/1990), medical schools have been required to include a course in human genetics as part of their medical curriculum. The duration of the course varies from 10 to 60 h from university to university. This modest attempt to initiate medical students is hoped to be only a start and is in no way an appropriate or adequate level of genetic teaching. The situation for biology students is better. Schools teach one complete course (over 150 h) of basic genetics, and offer other optional courses in specific areas: cytogenetics, population genetics and molecular biology.

There are no masters programmes for laboratory scientists although short courses in general cell biology and molecular genetics are regularly offered by different centres and universities. There are also doctoral courses in medicine and biology that include human and medical genetics. Training for genetic counsellors or genetic nurses does not exist and genetic counselling is almost exclusively carried out by medical personnel although counselling may sometimes be provided by non-medical scientists in some prenatal diagnosis clinics.

There are no official training programmes in clinical genetics, cytogenetics or molecular genetics. For certified medical doctors or non-medical graduates, the lack of recognition of the specialy impedes their access to a genetic unit/centre through the National Training Programme for Medical Specialists. Instead, training is unofficially provided in genetic units/centres with facilities in clinical genetics, cytogenetics and/or molecular genetics, most frequently by collaboration in research projects. There are no arrangements for inspecting training units/centres. The great overload on diagnostic services in health centres limits opportunities for research and training. Accreditation for genetic trainees does not exist. However, a certified degree following a course on medical genetics has been offered yearly since 1995 by the University of Alcala de Henares (Madrid) and the Department of Medical Genetics of the Hospital Ramon y Cajal (Madrid). The course is available to medical doctors with or without a medical specialty and the contents include basic concepts of human genetics, a broader approach to clinical genetics and its possible application to different fields of medicine.

There are no specific arrangements for linking clinical genetics, cytogenetics and molecular genetics in tertiary centres, although in practice this tends to occur. Research and education are usually included in these centres but are not required as a condition for recognition as a refer-

ence centre. Laboratory equipment is not, generally, a limiting factor for genetic services.

Dimension 5: Non-Harmful

Spain has one main professional organisation in human genetics: the Spanish Association of Human Genetics. Two additional organisations (Spanish Association of Prenatal Diagnosis and Spanish Association of Clinical Chemistry) also include professionals in human genetics. There is general agreement among human geneticists that genetic services must provide appropriate genetic counselling as well as diagnosis and that only medical geneticists, physicians, pharmacists and biologists having competence in genetics should have the authority to provide genetic testing. In practice, many units/centres perform cytogenetic and molecular analysis requested by professionals without genetic training and counselling cannot be guaranteed. There is no specific legislation to ensure the appropriateness of genetic procedures and the confidentiality of personal data. Quality assessment schemes for genetic services are practically non-existent. Only laboratories that perform neonatal screening work within a national quality assurance programme. This is controlled by the Spanish Committee on Inborn Errors of Metabolism of the Sociedad Española de Bioquímica Clínica y Patología Molecular. Seven biochemical genetic laboratories also collaborate with the European Network for the Evaluation and Improvement of Screening, Diagnosis and Treatment of Inherited Disorders of Metabolism, supported by the European Community. However, there are no organisations responsible for the maintenance of standards in clinical, cytogenetics or molecular diagnostics although in 1994 a Committee for Accreditation and Quality Control of Cytogenetic Laboratories was set up [7] and standardised criteria for accreditation and quality control have been introduced recently. Plans to develop quality standards for clinical and molecular genetics are presently in process.

Dimension 6: Effectiveness

Measures of the effectiveness of genetic services are related to the incidence of genetic disorders, medical reports on genetic counselling and testing, and medical and population knowledge on genetic services available to the community. Currently in Spain there are at least five registers of congenital malformations and inherited disorders and these have an important role in birth defect surveillance. These registers include a nationwide hospital-based case control study (The Spanish Collaborative Study on Congenital Malformations) and four population-based registers for the communities of Asturias, Vasc Country, Navarra, El Valles region and the city of Barcelona. The prevalence of DS in the newborn in Spain has decreased since 1986 [8] and similar trends have been observed for anencephaly and spina bifida with a decrease of 4.6 and 1.8 cases per 10,000 live births, respectively [8]. These findings are related to the availability of prenatal screening and diagnosis for these disorders, as well as the legal option of abortion.

Prevention of genetic disorders is, in most regions, widely accepted, as indicated by the increasing demand for genetic counselling and testing. The authors do not have data on the effectiveness of fetal ultrasonography to detect congenital defects. However, the number of cytogenetic analyses performed prenatally has significantly increased during the last 10 years. It can be estimated that approximately 15–20% of the total number of mothers at increased risk for DS (35 years of age and older) receive fetal karyotyping.

Prospective studies of individuals found to be genetically susceptible to breast cancer or polyposis coli have only recently been initiated and the authors do not have data to use as a measure of the effectiveness of genetic services in Spain.

Dimension 7: Consumer Satisfaction

There are no measures of consumer satisfaction, the rights of consumers to information, social acceptance of genetic services or their influence on policy makers. However, genetic services are widely accepted in our society as indicated by the increasing demand. Research in human genetics, and especially the Human Genome Project and its future applications to medicine, have been frequently discussed in the media. In fact, information overload on genetics has generated false expectations and perceived threats to consumers. This has facilitated open public discussion of scientific and ethical issues, contributing to improvements in the education of the population.

The number of genetic patient organisations in Spain has significantly increased in recent years. Although some are well organised and very active, they do not have resources to support research.

Publications Related to Genetic Services

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