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Genetic Services in France

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Country Background: Demography, Geography and Infrastructure

France has a population of about 58 million people, is 543,965 km^2 in size and has a population density of 106/km^2 (1993); 74% of the population live in urban areas. France is a republic, has a highly developed transport and communication network and is economically well developed. There is no official state religion.

The ethnic composition of the country is as follows: 93.6% of the population are French, Gypsies account for about 275,000 and 2.6% of the population are second-generation Arabic immigrants. Foreign nationals account for 6.4% of the population and include 1.4% from Portugal, 1.3% from Algeria, 0.9% Spain and 0.9% from Italy, Morocco, Tunisia, Croatia, Turkey, Senegal, Mali and other West African countries. There are two regions where foreign nationals with specific conditions (haemoglobinopathies) are concentrated: the Paris region and the Mediterranean Sea border, where African, Mediterranean and Caribbean people are living.

Health Service Setting

The French health care system is an attempt to reconcile solidarity and liberalism by means of a combination of collective financing and a public and private sector for the delivery of health care. The government has control of the health insurance schemes, it determines the level of doctors' fees and the prices of pharmaceuticals and investigation procedures. The 22 regions and 95 departments are all involved to a certain extent in financing and delivering health care. There is a national umbrella organisation for the public health insurance scheme, which is responsible for an equitable availability of health care and general budget control; 99% of the population is protected by this system. The insurance system is based on direct payment of the physician by the patient and reimbursement of part of the charges. Health costs resulting from copayment for almost 80% of the population are covered by mutual societies.

Primary medical care is provided both by GPs and independent specialists working from their own premises. In most cases, the GP is an independent doctor practising alone. Patients are free to choose their physician. Thus, people are not registered with a doctor. Since referrals by a GP to specialist care are not required, GPs do not have a gate-keeping role.

The French hospital services consist of different types of establishment; over two-thirds are public hospitals with the remainder being private for-profit hospitals and private non-profit hospitals, with very few religious hospitals. It should be noted that the spread of doctors and health care services varies greatly. Supply in the Paris region and along the Mediterranean Sea is very much higher than in rural areas.

GPs represent 54% of all physicians and do the follow-up of 40% of pregnancies during the two first trimesters. The first antenatal visit takes place during the first trimester in the vast majority of pregnancies. The follow-up of the pregnancy is done exclusively in private practice in 42% of cases, exclusively in public hospitals in 37% of cases, the others being followed up by both systems. Half of the maternity hospitals are public and half are private.

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Table 1. Number of medical genetics staff engaged in genetic service provision in France

Number of centres	Population served	Genetically trained physicians	Genetic physicians in training	Cyto-geneticists	Molecular geneticists	Genetic nurses/ counsellors
NA	58 million	125	30/year	200	100	0

NA = Not yet applicable, as medical genetics centres will be defined by application of the new law in 1997 but are likely to remain local networks rather than centres.

History of Medical Genetics

Medical genetics developed both from cytogenetics and paediatrics in the 1960s, mainly in public hospitals. Most clinics were opened in other departments as genetics was not a recognised specialty, but some were attached to cytogenetics laboratories or to neurology departments, depending upon the interest in genetics of motivated clinicians. Until now, the development of medical genetics, apart from the pricing structure, has not been planned at a central level, which explains why there are very few genetics centres offering all the services in one place.

Prenatal diagnosis of chromosomal anomalies started in 1971 in Paris and developed slowly, with 5 laboratories in 1976, 11 in 1980 and 70 in 1997. The constitutional karyotype was reimbursed at a very low price, to prevent the development of private for-profit laboratories. Price readjustment took place in 1992.

Activities in biochemical genetics and molecular genetics were developed mainly by research teams and only from 1996 recognised as routine investigations reimbursable by national health care insurance, having previously been funded by research grants or public hospitals.

Because of the increasing activity in medical genetics, in 1980 a contract was signed at the national level between the national health care insurance and an association of professionals (Association Française pour le Dépistage et la Prévention des Handicaps de l'Enfant, AFDPHE). Through this contract, all the investigations around prenatal diagnosis and all the biochemical and molecular genetics tests were paid directly by the association to the laboratories, being free of charge for the patients. The definition of the tests which could be charged in this way was negotiated on an annual basis. In 1992, most of the cytogenetic tests being considered as routine practice, the price of their reimbursement was defined and they were no longer paid through the association but directly through health insurance. The same process occurred in 1996 for molecular and biochemical tests.

In 1995, medical genetics was recognised as a specialty. Before that, genetics was considered as a sub-specialty from 1983, although a professorial appointment in medical genetics was created at a national level, potentially for all universities, as early as 1981.

Since 1988, laboratories have to be authorised by the Ministry of Health to perform any prenatal diagnostic tests with, from 1995, the non-observance of the regime of authorisation punishable by a prison sentence. Laboratories have to apply separately for each type of activity, i.e. cytogenetics, molecular biology, biochemistry, serum screening. They are authorised for 5 years and have to produce an annual report on their activity. The advisory committee to the minister for authorisation is composed of qualified professionals, of representatives of professional organisations, lay associations and different bodies such as the National Advisory Ethics Committee and INSERM, and representatives of the Ministries of Health and Justice.

Dimension 1: Availability

In 1996, French genetics services were organized as shown in table 1.

Fifty-three laboratories (around 1 per million people) currently perform molecular biology tests for genetic conditions. They offer tests for 171 different genetic conditions, the most common ones, offered by more than 10 different laboratories, being for Duchenne muscular dystrophy, haemophilia, cystic fibrosis, myotonic dystrophy, and fragile X syndrome. From this group of 53 laboratories, 37 were authorised to carry out prenatal diagnostic tests.

There are 98 cytogenetics laboratories (around 1 per half million people) and 70 of them are authorised to car-

ry out prenatal activity. There are 200 cytogeneticists. The average number of karyotypes/cytogeneticist is 1,000 and the average number of karyotypes/technician is 300. FISH is theoretically available for DiGeorge, Williams and Prader-Willi syndromes in most laboratories. There are also 31 biochemistry laboratories which have prenatal authorisation, performing tests for genetic diseases.

In the whole of France there are 62 genetic clinics (1 per million people) with 125 clinical geneticists seeing patients. Some of these clinics have a multispecialty approach for specific conditions such as Huntington disease, neurofibromatosis, Marfan syndrome, cystic fibrosis and Duchenne muscular dystrophy. There are no genetic counsellors or genetic nurses involved in these clinics.

Cancer genetics is established as a separate network, including 33 clinics where 54 oncogeneticists are seeing patients.

For historical reasons, most of the facilities are not coordinated in identifiable genetics centres. In all large towns, all the facilities are available but in different hospital departments and even in different hospitals. This is why the bioethics law passed in 1994 mentions the multispecialty/multidisciplinary centres as the structure for genetics services in the future, which makes it obligatory that geneticists reorganise their services. Starting in 1997, prenatal genetics centres will have to be authorised.

There are two genetic registers in France, one dedicated to cystic fibrosis and one to sickle cell anaemia. There are also four registers of birth defects including chromosomal anomalies and genetic syndromes recognisable at birth. These birth defects registries are population based and survey 25% of the births in France (Paris, Bas-Rhin, Bouches-du-Rhône, Centre-Est-Auvergne).

Population screening is performed only at the neonatal period for PKU, congenital adrenal hyperplasia and hypothyroidism. A national experimental programme for screening for cystic fibrosis at birth ended in 1992 with information on 800,000 births (although this programme is still active in some regions). Neonatal screening for sickle cell anaemia is performed on children at risk because of their ethnic origin (100,000 tests/year).

Dimension 2: Access

There is no shortage in genetic services as regards the number of diagnostic laboratories, which are available in every region, but genetic clinics are very limited and some regions lack clinical services.

Social security in France insures reimbursement for medical visits and biological examinations, according to a preset scale. Reimbursable activities are listed in a 'schedule of medical activities' as is the rate of reimbursement. At the present time, clinical work is considered equivalent to a typical medical consultation, with the reimbursement rate depending on the status of the physician-geneticist, a calculation which does not take into consideration the particular length of the genetic consultations.

The biological work mostly concerns karyotyping and molecular biology. There is no restriction in accessing karyotyping during the postnatal period. Fetal karyotyping is compensated within the following categories: if the mother is at least 38 years old, if an anomaly is found by ultrasonography, if a chromosomal abnormality exists in a sib, if one of the parents is a carrier of a balanced chromosomal rearrangement, and if the value of maternal serum markers indicated a risk equal or greater than 1:250. Molecular biology work has been included in the rate schedules since 1996.

The uptake rate of genetic tests is known only for prenatal diagnosis. In 1994, 60% of eligible pregnant women 38 years old and over had fetal karyotyping performed and the uptake of serum marker screening was 20% of all pregnant women in 1995. Various surveys have shown that social inequalities in accessing genetic services are the rule. There are also large regional differences, with uptake in Paris twice the rate of the lowest region.

Dimension 3: Life Sustaining

The life expectancy of women and men is 81.5 and 73.3 years, respectively. Infant mortality is 4.9 per thousand live births. The birth rate is 12.3/1,000 and the fertility rate, 1.65 per woman; 67% of the mothers are of French origin but only 3% of all pregnant women do not speak French.

Dimension 4: State of the Art

Professionals in Medical Genetics

Medical genetics only achieved formal recognition in university faculties when legislation was passed on April 15, 1983. Genetics is taught to undergraduate medical students in the first year (approximately 30 h), sometimes by instructors from the science faculty. Eight of the 37 faculties of medicine have no professor of medical genetics. Masters programmes in cytogenetics or in human

genetics were the only higher degrees available until 1988. From legislation passed on April 29, 1988, two diplomas were created: a diploma in molecular biology and a diploma in human cytogenetics. The latter was only available to those already specialists in medical biology, gynaecology-obstetrics, haematology, internal medicine, oncology or paediatrics.

At the request of the 'Conseil National de l'Ordre de Médecins' which is in charge of the regulation of the medical profession, including the recognition of specialities and competencies, a competency certificate in medical genetics was created by law on February 10, 1981. Review boards were only legally established on July 30, 1990 and physicians were then able to qualify in medical genetics as a subspecialty from 1990.

The specialty of medical genetics was created in 1995. It was proposed that 30 geneticists should be trained every year, bringing the total number of geneticists in France to 400 by 2005. This increase should be concurrent with an increase in geneticists working as medical university lecturers, professors and hospital physicians. The funding of these positions is not yet clear.

There is no training in genetics for nurses, midwives and psychologists and the profession of genetic counsellors does not exist. This is due to opposition of medical geneticists to the principle of having non-medically-trained health professionals advising patients in genetic clinics. There is a lack of trained cytogeneticists and research in cytogenetics, due to the attractiveness of molecular genetics for young scientists and physicians.

Dimension 5: Non-Harmful

No professional guidelines have been produced about teaching, training, practising and quality control by the geneticists themselves. The public guidelines already published were written by the National Advisory Committee on Bioethics, and by the National College of Gynaecologists and Obstetricians. The topics covered by these guidelines are prenatal diagnosis, preimplantation diagnosis and predictive testing for late-onset diseases.

There is no quality assessment network in cytogenetics, molecular biology or genetic biochemistry. Half of the laboratories performing serum marker screening belong to a quality control network.

A bioethics law was passed on July 29, 1994, directed at regulating some aspects of the genetics services. The main paragraphs of this law relevant to genetic services are the following.

(1) Prenatal diagnosis must conform to accepted medical practices. It should be preceded by genetic counselling.
(2) Cytogenetic and biological tests leading to prenatal diagnosis are only permitted under conditions outlined by state council order, and only in public health establishments and authorised laboratories.
(3) Genetic counselling preceding prenatal diagnosis is intended to inform the couple of the reasons for examination, the reasons for sampling and its risks, as well as the possible consequences of the diagnosis.
(4) A state of council order defines the conditions, in the patient's interest, for the use of genetic testing for medical purposes.
(5) The Minister of Health may, by decree, determine the proper practice and technical rules for genetic testing and, if necessary, the mode of medical follow-up.

Since May 1995, all laboratories involved in prenatal diagnosis have to apply to the Ministry of Health to be authorised for their specific activity. There are four categories: cytogenetics, molecular biology, biochemistry and serum marker screening. Authorisation is given for 5 years, and an activity report is due every year. This should contribute to the improvement of the services, if the law is applied, which means if non-authorised laboratories are obliged to stop their activity. The same authorisation procedure applies to preimplantation diagnosis and predictive testing.

Dimensions 6 and 7: Effectiveness and Consumer Satisfaction

There is no published evaluation of the effectiveness of medical genetics in reaching its goals. Some evaluation has been published in the field of prenatal diagnosis. As far as fetal karyotyping is concerned, 60% of all women 38 years and older were able to have a prenatal test in 1994. Under current conditions, it appears that the maximum percentage of eligible women requesting a prenatal diagnosis would be 75–80%. In 1994, 50, 000 fetal karyotypes were performed, 56.3% for advanced maternal age, 20.7% because of abnormal findings at ultrasound, and 9.1% after a serum marker screening test. At the national level, 49% of trisomy 21 were terminated.

As far as cancer genetics is concerned, a recent survey indicates that a great majority of consultees received the necessary information, with a minor percentage considering the consultations too technical.

Many patient organisations exist in France. The largest is the l'Association Francaise contre les Myopathies (AFM) which supports scientific research and diffuses information to patients and physicians. In most of these

associations medical geneticists are involved on the Board.

Information on genetic services in France is accessible via internet (http://www.infobiogen.fr/services/orphanet/), and a list of the lay associations in genetics is available on internet (http:/www.infobiogen.fr/agora/associations/). ORPHANET includes information on the conditions, on research laboratories working on these conditions, on diagnostic laboratories and specialized clinics. This server is financially supported by the Ministry of Health and INSERM. In 1997, it will be extended to all rare disorders. There is also a toll-free telephone line delivering information on genetic diseases and services to both patients and physicians which opened in 1995, financially supported by the Ministry of Health and the AFM.

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Validator

This chapter has been validated by the authors.