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Genetic Services in Italy

Franca Dagna Bricarelli[a]
Bruno Dallapiccola[b]
with the co-operation of *Renzo Provedel*[c] and *Giovanni Romeo*[d]

[a] EO Ospedali Galliera, Genoa,
[b] Università 'Tor Vergata', Roma,
[c] AIRH, Genoa,
[d] Istituto G. Gaslini, Genoa, Italy

Country Background: Demography, Geography and Infrastructure

The population of Italy in 1995 was 57,332,996 with a total of 526,064 newborns. The population is highly heterogeneous as a result of many migrations through the centuries. The high frequency of thalassaemia and haemoglobinopathy heterozygotes is particularly noteworthy in the populations originating from South Italy, Sardinia, Sicily, Po River delta, and the Ferrara country.

Health Service Setting

Health care expenditure is about 5% of GNP. The 'challenges' to health services result from their current bureaucratic and weak structure and historical precedents. Based on the most recent reform law of the National Health System (SSN, 1992), the situation is characterised by great autonomy, both organisational and financial, at the level of the 20 regions to which the National Ministry of Health assigns budgets based on population numbers. In addition, the Ministry publishes basic guidelines, standardises the level of assistance, defines regional responsibilities, promotes partnerships with universities, defines standard fees for hospitals and agreed private bodies, and defines the personnel employment criteria in the public and certified private bodies. At the regional level, health care is organised by the Aziende Sanitarie Locali including hospitals and territory services (in Italian: poliambulatori). A number of large and highly specialised hospitals are managed as 'enterprises' with strong economic and financial autonomy. Thirty-two hospitals which are classified as 'Istituti di Ricovero e Cura a Carattere Scientifico' receive special funds for research from the Ministry of Health. The budgets are allocated at a regional level, according to the specific local services and the volume of activities.

Dimension 1: Availability

The provision of genetic care in Italy does not foresee formal referrals at different levels of expertise and of laboratory competencies. However, several centres of medical genetics at regional or national level are informally recognised as points of reference for specific genetic disorders.

Primary genetic advice is usually provided in Italy by paediatricians, obstetricians and more rarely by GPs. They refer their patients to established institutions where one or more genetic clinics are in charge of the out-patient service. More often patients without a diagnosis are admitted to hospital wards. A representative example of this informal organisation is provided by the Liguria region which provides for 1.5 million inhabitants the highest number of genetic centres. Due to this tradition in genetic health care, a regional Department of Medical Genetics is now being organised in Liguria. This department, the only one in Italy, should provide the framework and the technical expertise for the co-ordination of genetic services in the region (including screening programmes, prenatal diagnosis, molecular diagnosis and genetic clinics).

There is no census to evaluate the number of medical geneticists in Italy and there is no national guideline to regulate their distribution in the country. About 400 med-

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

F. Dagna Bricarelli
EO Osped Galliera
Genetica Umana
I–16128 Genova (Italy)

ical doctors, physicians and biologists are specialised as medical geneticists. However, only a minority work as medical geneticists in public services. The coverage of local needs is planned by the regional authorities.

National Network of Genetics Centres

Regarding the organisation of the services, there are valid structures and professional competencies: universities, scientific institutes, hospitals at international-standard levels of service. Services are not homogeneous throughout Italy due to different capabilities and funding in each region. A general evaluation of the Italian scenario has been presented in *Genetic Diseases: Guide to Diagnostic Services, Scientific Associations and Charities* published in 1994 and sponsored by the different Italian Scientific Associations, AIRH (a national charity) and the Ministry of Health. This guide, which is updated on line, has been diffused by internet (www.Genet.it), and also distributed to physicians.

Regional (Population-Based) Organisation of Genetic Services

The distribution of genetic laboratories and clinical services in the Italian territory is not uniform but is related to local needs (e.g. populations, risks, local facilities, specific interest of the local physicians). The ratio of services/population is high due to an inadequate planning rather than to a high service level.

Under One Roof (Combined Clinical and Laboratory Services)

With the exception of the recently instituted Department of Genetics of the Liguria region (not yet operative), there are few examples of integrated systems for the diagnosis and prevention of genetic disorders.

Molecular Diagnosis Available in Major Centres

In each region, the common genetic diseases are diagnosed by a molecular approach; the rare diseases are analysed by major reference centres. Diagnosis of most disorders is available in Italy. The focal points are usually the laboratories. The clinical services are associated with them. About 50 clinical services with annexed laboratory facilities are distributed in different regions. Biochemical genetic networks are organised along the same lines.

Genetic Nurses/Genetic Associates

There is no tradition in this field. There are some local initiatives of training and employment but no official and certified courses.

Genetic Family Registers

The National Institute of Health Care ('Istituto Superiore di Sanità') provides yearly statistics on the most important congenital malformations in the country. Genetic family registers are kept by the laboratories based on the referred patients. In some cases, a laboratory becomes a national reference for a specific disease.

Cytogenetic Diagnosis of Common Genetic Disorders

In each region there is at least one laboratory with the knowledge and capability to diagnose the main disorders. FISH testing is available in the major cytogenetic laboratories.

Population Screening

Population screening is regulated by regional laws. All regions provide neonatal screening programmes for hypothyroidism and PKU. Screening and prevention of the commonest pathologies including thalassaemia, galactosaemia, and a few other disorders are performed according to the different regional programmes.

Pregnancy

A national law (1995) gives a detailed list of examinations for pregnancy monitoring to be provided free of charge. Genetic counselling is performed in about 90% of the hospitals and university centres. Genetic counselling preceding prenatal diagnosis is available in all public services, but only in a minority of private ones. It is common for most centres to ask the patients to sign an 'informed consent' form prior to an invasive examination. Ultrasound screening is currently performed in about 85% of pregnant women, usually in the second trimester. Chorionic villus sampling (CVS) is currently performed for DNA analyses. CVS for cytogenetic prenatal diagnoses is used in less than 5% of women requiring foetal karyotyping.

Professional Organisations

There have been professional genetic organisations since 1975. Currently there are three major professional Societies: AIGM – Medical Genetics, AICM – Medical Cytogenetics, SISMME – Inborn Errors of Metabolism. These Societies became associated in 1980 to form the FISME – Italian Federation for the Study of Inherited Diseases, and are now considering the creation of a single society – SIGU – Italian Society of Human Genetics. An additional society, AGI – the Italian Association of Genetics – is concerned with general genetics, excluding medical genetics.

Cancer Genetics

A network of cancer genetic clinics and laboratories is being organised with funding and co-ordination from a private charity (Associazione Italiana per la Ricerca sul Cancro, AIRC). The programme is being developed at present in two main directions: diagnosis and prevention of colorectal cancer and diagnosis and prevention of breast cancer.

Dimension 2: Access

Funding for genetic services comes from the National Health Service. Fetal and postnatal karyotyping, molecular diagnostics and genetic counselling are provided almost free of charge by the public services. The limitations of this system are apparent in regions (especially in the south of Italy) where public services cannot cope with the demand. In these regions, private laboratories and services are generally stronger than in the north, and they are often organised to replace the public service completely. The 1995 national laboratories census carried out by AICM (Italian Association of Medical Cytogenetics) found that 22 out of 116 laboratories were private (about 20%) but that about 30% of examinations were performed privately. However, 50% of prenatal diagnoses were performed privately, mainly for 'low-risk' pregnancies with maternal age less than 35 years. In contrast, less than 5% of molecular diagnoses are carried out in private centres, which include mainly screening for the most common form of β-thalassaemia, cystic fibrosis mutations and paternity testing. In the south of the country the number of examinations is lower than the national average and they are performed more in the private structures.

Religion is not a major obstacle to access to services. This is clearly shown by the success of thalassaemia screening and prenatal diagnosis in Sardinia. In addition, prenatal diagnosis is currently performed in some Catholic institutions. The abortion law allows termination for fetal abnormality after the first trimester based on the recognition of psychological and physical damage to the mother certified by a medical specialist.

Dimension 3: Life Sustaining

Infant mortality is 6.4 per 1,000, male life expectancy 74.9 years and female life expectancy 81.4 years (1995).

Dimension 4: State of the Art

There are no published recommendations for teaching genetics to medical students, training medical geneticists, laboratory geneticists, or genetic nurses/associates.

Postgraduate university training in medical genetics is provided by two types of course: (1) PhD programmes (3–4 years) and (2) nine postgraduate schools of medical genetics (specialisation of 4 years). These lead to a qualification as a 'specialist'. Both physicians and biologists can attend these courses which provide the only official qualifications for and recruitment to clinical and laboratory genetic services.

Overall about 70 students are admitted each year to the schools of specialisation in medical genetics but only some of them are supported by a fellowship. There are an average of 5–15 fellowships available each year in each school. In each school, 2–3 fellowships are provided by the Ministry of Health (or National Health Service) and are reserved for MDs. Other fellowships for biologists (1–2 per school) are awarded by the Ministry of University and Research. Access to each school of specialisation is regulated by local competition. There is no nationwide certification of competence for geneticists, and the qualifications from the schools of specialisation are the only proof of training in genetics, both for biologists and MDs.

An important feature of the Department of Medical Genetics, currently being established in Liguria, is the development of educational programmes in genetics for GPs. Training programmes for genetic nurses do not yet exist in Italy. Although there are recommendations for general laboratories, there are none for minimum laboratory equipment in genetics laboratories. Research in Italy is active and is supported by universities, the Health Ministry, international grants, and by private grants (e.g. Telethon).

Dimension 5: Non-Harmful

In Italy there are no laws regulating genetic research and in general there are no constraints on scientific development. However, a directive from the Health Ministry is expected to be issued in 1997 about limitations to be imposed on embryo research and animal cloning. There are no published audit reports relating to genetic services, no inspection/approval of training centres, no accreditations/certification of specialists and no laboratory quality assessment. The Italian Medical Cytogenetics and Medi-

cal Genetics Associations prepared their guidelines for genetic analyses, but there are no formal rules enforced by public agencies. The National Ethical Council is publishing audit recommendations on different subjects including prenatal diagnosis and gene therapy. Private organisations, for example the 'Tribunal of patients', have been set up in recent years to enforce a policy on the rights of patients and are providing guidelines for 'consumers' protection.

Dimension 6: Effectiveness

There are no specific publications.

Dimension 7: Consumer Satisfaction

There are no published reports on consumer satisfaction although there are regular public debates on genetic issues. There are many active patient organisations but only a few of them are influential, including notably the Cystic Fibrosis Association, the Down Syndrome Association and the Muscular Dystrophy Association. There is no 'umbrella' patient organisation.

Public awareness of genetics has increased in recent years, mainly because of mass media coverage. In primary and secondary schools, many initiatives aim to improve the basic knowledge of the students and are leading to a better understanding of genetic principles and diseases.

Publications Related to Genetic Services

1 Guida ai Servizi 1994 Diagnostici e alle Associazioni – Malattie Genetiche (by the National Health Deparment and AIRH).
2 Linee Guida per la Diagnostica Citogenetica 1995, by Consensus Conference – AICM.
3 All above information available on the internet: www.genet.it.

Italian Society for Human Genetics

In 1997 the following societies are active:
(1) AIGM (Italian Association of Medical Genetics)
(2) AICM (Italian Association of Medical Cytogenetics)
(3) SISMME (Italian Association of Inborn Errors of Metabolism)

At the end of 1997, the fusion of the AIGM and AICM into a new society called SIGU (Italian Society of Human Genetics) is expected.

Validator

Prof. B. Dallapiccola, MD
President, AIGM
Cattedra di Genetica Medica
Università degli studi di Roma 'Tor Vergata'
Via di Tor Vergata 135
I–00133 Roma (Italy)

'The Associazione Italiana di Genetic Medica (AIGM) has evaluated and approved the chapter prepared by F. Dagna Bricarelli, B. Dallapiccola, with the collaboration of G. Romeo and R. Provedel, for CAGSE, concerning the state-of-art of Genetic Services in Italy.'