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Portugal: The practice of medical genetics in Portugal

The study of genetic diseases started in Portugal with the diagnosis and family evaluation of adult-onset diseases, in 1939 for amyloid polyneuropathy and in 1978 for Machado-Joseph disease.

In the 1970s several physicians, most of them pediatricians, developed a special interest in the diagnosis and prevention of congenital malformations and genetic counseling. Training was mainly done abroad. Clinical and cytogenetics services were first provided at the Faculty of Medicine of Porto, and a genetics unit with its own cytogenetics laboratory was created in 1975 in the Department of Pediatrics of the Hospital de Santa Maria, Lisbon.[1] In the 1980s the introduction of a national neonatal screening program for phenylketonuria and hypothyroidism was one of the cornerstones of the Instituto de Genetic Medical Jacinto de Magalhaes, in Port, which is one of the main providers of genetic services in Portugal, in both the clinical and laboratory areas. In Lisbon, these two sectors have developed independently. The laboratory facilities are mainly at the Centro de Genetica Humana, at the Instituto Nacional de Saude Ricardo Jorge, while the clinical care providers are the departments of genetics at the Hospital de Santa Maria, Hospital Egas Moniz, and Hospital Da Estefania, all of which also have cytogenetics laboratories.

Also in the 1980s, the widespread use of ultrasonography in obstetrics and the beginning of cytogenetic prenatal diagnosis drew the attention of obstetricians to this area. This was especially so after the passage of the law permitting termination of pregnancy in 1984. The Prenatal Diagnosis Association was created in 1995.

The 1990s saw a significant expansion in the number of molecular diagnostic laboratories as well as the establishment of private genetics laboratories. Prenatal biochemical screening for trisomy 21 after the first trimester also became widespread. Two other genetic services were created, one in Vila Real and the other in Coimbra.

There is now a public and private network covering clinical genetics, cytogenetics, and molecular genetics. Collaboration with international reference units is required for the study of most rare conditions.

The Ministry of Health passed legislation on molecular genetics testing (diagnostic, including prenatal diagnosis, carrier detection, and presymptomatic diagnosis) and prenatal diagnosis in 1997.

Portuguese groups are also involved at both the national and international levels in human genetics research. Most relevant are the studies being performed in two autosomal dominant late-onset diseases particularly prevalent in our population—amyloid polyneuropathy and Machado-Joseph disease.

Teaching genetics at medical schools

The teaching of medical genetics in Portugal started in 1970 at the Faculty of Medicine of Porto, initially under the "umbrella" of medical pathology and, since 1981, as an independent subject.[2] Other medical schools soon adopted similar teaching programs. In the last half of the 1990s, some medical schools also offered clinical genetics courses during the clinical teaching years, while keeping the genetics courses at the preclinical teaching level.

Training in medical genetics

In 1983 the Portuguese medical board (Ordem dos Medicos) recognized medical genetics as a specialized area (but not yet as a specialty). A formal 2-year training program in medical genetics has been available since 1986 for specialists in a few other areas.

In 1999, medical genetics was officially accepted as a medical specialty and the College of Medical Genetics was created. The training program has a duration of 5 years, as is usual in Europe.

Medical scientists working in public institutions are required to complete a 3-year postgraduate training course covering cytogenetics, biochemical genetics, and molecular genetics. To cover other sectors, the College of Human Biology and Health of the recently created Board of Biologists (Ordem dos Biologos) has drawn up regulations pertaining to professional prerequisites and practice.

Portuguese Society of Genetics

The Portuguese Society of Genetics, founded in 1973, includes members from all fields of genetics.[3] It regularly publishes the scientific journal *Broteria Genetica*.

Portuguese Society of Human Genetics

The rapid development of human genetics (clinical care, laboratory diagnostic facilities, and research) prompted physicians, researchers, and laboratory scientists working in this area to create the Portuguese Society of Human Genetics in 1996. This society has so far organized four annual scientific meetings and hosted in 1998 the 30th annual meeting of the European Society of Human Genetics. It regularly publishes a newsletter, which is available to its 351 members, and has appointed an Ethics Committee. In collaboration with The British Council, the Ethics Committee organized the international conference "Issues in Human GenEthics" held in Lisbon in June 2000.

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Greece: The Hellenic Association of Medical Geneticists

The Hellenic Association of Medical Geneticists (HAMG) was founded in 1982 by a group of scientists specializing in genetics. The founding members were 21 doctors, mainly pediatricians and biologists working as senior staff in genetic units of university departments and major hospitals in Greece. At present there are 160 active members of HAMG, and all medical specialties are represented, including biology, biochemistry, and dentistry; in 1988 the society had only 59 members.

The principal goals of HAMG are the establishment and promotion of genetics in Greece, research on genetic diseases, and dissemination of information to the public on available genetic services for the prevention of inherited disorders. Another basic objective is the scientific recognition and professional consolidation of geneticists in Greece.

The association's activities have focused on the development of a close collaboration and exchange of opinions among all medical doctors and other scientists who are involved in genetics in Greece. It is worth mentioning that the effort for close collaboration with other Balkan countries was fruitful and resulted in the establishment of a biannual Balkan Medical Genetics Congress. The first and the third Balkan medical congresses were held in 1994 and 1998 in Thessaloniki, Greece, with the participation of geneticists from Europe and the USA; the second meeting was held in Istanbul, Turkey; and the fourth was held in 2000 in NoviSad, Yugoslavia. During the third Balkan Congress in Thessaloniki, the establishment of a Balkan Medical Genetics Association was thoroughly discussed and decided upon.

Meeting seminars and conferences sponsored by HAMG have been held in Athens, Thessaloniki, Ioannina, Corfu, and Crete, with distinguished Greek and foreign lecturers.

As the only representative of the Greek geneticists, HAMG has applied to the Health Ministry for the foundation of the necessary Medical Genetic Centers, aiming at the improvement of genetic services in Greece. Also, following the universal demand for the establishment of a medical genetics specialty, HAMG has focused its activity in this direction. We hope that the recent establishment of medical genetics as an independent department of the University of Athens will promote these efforts.

One year ago, HAMG began publication of the first Greek journal on genetics; the *Hellenic Journal of Human Genetics* is intended not only for the members of HAMG, but also for every scientist interested in genetics.

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