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## Ordem dos Medicos

### Report

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To: Lucia Mestre  
Subject: Information for the European Commission on the Medical Genetics Specialty  
Date: 14 September 2009

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As part of the work of the Committee on the recognition of qualifications provided for by Directive 2005/36/EC, the European Commission requested a brief summary of the specialty of Medical Genetics.

Order No 148 of 2 March 2001 setting up the professional field of medical genetics provides the basis for the following:

#### SUMMARY OF THE TRAINING IN MEDICAL GENETICS

Access to training in medical genetics requires prior basic medical training and is gained via national competitive entry to medical traineeships. Such competitive entry is common to all the medical specialties and candidates are ranked in order to choose their preference from among the various specialties available every year. Training in medical genetics is regulated by Order No 148 of 2 March 2001 which lays down the duration, structure, training establishments, objectives and assessment for the training programme.

This breaks down as follows:

NAME OF THE QUALIFICATION: MEDICAL GENETICS

DURATION – 60 MONTHS

- basic clinical training (minimum 12 to 15 months) practical training period in prenatal diagnostics (3 months); 2 periods of practical training in paediatrics (of 3 months each) in neonatology, developmental paediatrics; practical training period in internal medicine or other adult specialty (3 months).
- laboratory practice (minimum 9 to 24 months): practical training period in cytogenetics (3 months), practical training period in foetal diseases (3 months) and optional training period (3 months) in one of the following: cytogenetics, biochemical genetics or biochemical and molecular genetics
- specific clinical training (minimum 24 to 36 months) in clinical genetics (including genetic counselling, predictive genetics, prenatal diagnosis, dysmorphology, metabolic disorders, adult genetic disorders)

- optional training periods (12 months) consisting of two 6-month periods to reinforce the laboratory component, clinical practice and/or provide greater opportunity to pursue more clinical or laboratory hours according to the future career profile of the trainee geneticist.

It is possible to replace the optional training periods with extended basic clinical training (instead of the optional laboratory practical training), extended laboratory practical training (instead of the optional training periods) or with extended specific clinical training (instead of the optional training periods).

The current programme of specialised training in medical genetics is perfectly compatible with recognition of qualifications of 4 or 5 years of specialisation, given that the traineeship programme in Portugal includes a first year consisting of basic clinical training (required for all medical practitioners), and specific training takes place over the subsequent 4 years. This is why there is no difficulty in recognising specialised training in medical genetics obtained in other countries with a minimum of 4 years of specialised training.

## TRAINING SITES

Portuguese law requires training sites to be approved as suitable for the provision of medical specialty training. Approval of institutions and health departments as suitable for medical training, as sites and in terms of the training offered, is by way of an order of the Ministry of Health subject to the technical opinion of the Ordem dos Médicos (see Article 11 of Decree-Law No 203 of 18 August 2004 adopting the legal framework for post-doctoral medical training with a view to specialising, and laying down the general procedural rules therefor).

In particular, with regard to training in medical genetics, Order No 148 of 2 March 2001 provides:

Practical training periods in cytogenetics: in cytogenetics laboratories belonging to medical genetics departments or institutions; the optional 6-month practical training periods may nevertheless be carried out in other approved laboratories which are dedicated to human chromosome disorders;

Practical training periods in biochemical and molecular genetics: in molecular genetics laboratories belonging to medical genetics departments or institutions or in other laboratories dedicated to molecular diagnosis of genetic disorders;

Practical training period in foetal diseases: in pathological anatomy departments recognised in the field of foetal diseases and approved for this kind of training;

Optional practical training period in biochemical genetics: in biochemical genetics laboratories, metabolism and/or enzymology units belonging to medical genetics departments or institutions or in other laboratories dedicated to genetic disorders;

Practical training period in prenatal diagnosis — in obstetrics departments with a level II or III prenatal diagnosis centre;

Practical training periods in paediatrics: in paediatrics departments with a neonatal unit and offering developmental paediatrics or metabolic disorders;

Practical training periods in internal medicine or other adult medical specialty: in a large medical department in a hospital or in departments which must not be paediatric but must essentially not be surgical or laboratory departments (internal medicine, neurology, endocrinology, cardiology, ophthalmology, orthopaedics, oncology and so on) where these are separate departments, provided a minimum of three months is spent in each of those departments;

Optional practical training period in epidemiology: in a department or institution approved for this kind of training, with public health programmes preferably relating to genetic disorders.

Optional practical training period in health statistics and ICT: in a department or institution approved for this kind of training;

Practical training periods in clinical genetics: in medical genetics departments or institutions, for optional practical training periods, or in departments or institutions which operate specialist or thematic clinics for diagnosis, treatment and follow-up of genetic disorders and/or for predictive tests and genetic counselling, provided a minimum of three months is spent in each of those departments or institutions.

## PERFORMANCE AND KNOWLEDGE OBJECTIVES

Order No 148 of 2 March 2001 also lays down the objectives for training in Medical Genetics, in particular the specific objectives for each practical period of the training.

The general objectives for specialised training in human genetics are therefore as follows:

- Training of clinical or laboratory geneticists to be able to offer prevention, diagnosis (clinical or laboratory) and appropriate therapy for patients suffering from genetic complaints and genetic counselling for such patients and their relatives, discussing all the available options, including as regards reproduction;
- Guidance for trainees to ensure the quality of their laboratory and clinical work, that they understand the psychological, family and social implications of genetic disorders and on the fundamental ethical and deontological principles governing clinical and laboratory genetic medical practice.
- Awareness of the importance of collaboration between geneticists responsible for laboratory tests and clinicians and of the need to deal proficiently and sensitively with doctors from other specialisms and with non-medical professionals involved in the care of patients suffering from a genetic disorder and their families in order to provide the most appropriate support to the patient;
- Evaluation of the constant developments in the field, in particular in such a vast area which is expanding so rapidly, in order to fill the lacunae in their knowledge by resorting to the appropriate resources;

Performance objectives are laid down for each practical training period at even intervals through the specialist training in human genetics:

| <b>Practical training period</b>   | <b>Performance objectives</b>  | <b>Knowledge objectives</b>  |
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| Cytogenetics                       | Practice the more usual techniques of cytogenetics, including karyotyping of lymphocytes from peripheral blood, amniocytes and chorionic villus, fibroblasts, bone marrow cells and the various chromosome banding methods and fluorescence in situ hybridization (FISH);  | Knowledge of the basic foundations of each technique and their appropriateness, knowing the main cytogenetic tests, in cases of chromosome disorders, leukaemia and solid tumours, how to interpret the results and their limitations; |
| Biochemical and molecular genetics | Familiarisation with the basic techniques of molecular biology, including extraction and quantification of DNA, the use of restriction enzymes and RFLP (restriction fragment length polymorphisms) southern blotting, PCR (polymerase chain reaction) and SSCP (single strand conformational polymorphism) in the study of normal variations and detection of pathogenic mutations; | Knowledge of the basic foundations of each technique and their appropriateness, knowing the main molecular tests, in cases of genetic disorders, how to interpret the results and their limitations;                                   |

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| foetal diseases                 | Familiarisation with the more common techniques for macro- and microscopic study of human anatomical pathology and, in particular foetal and placental pathology; active participation in post-mortem examinations of foetuses and neonatals; observe and correctly note (by inspection and measurement, photography, x-ray and anatomical pathology) any anomalies found in materials from aborted foetuses and stillbirths and as the result of examining foetal membrane; | Recognition of the main congenital anomalies and multiple malformation syndromes and the main placental pathologies, identification of their causes (whether genetic or acquired) and methods of prevention;  |
| Optional – biochemical genetics | Exposure to the main basic biochemical techniques, including chromatography, HPLC (high performance liquid chromatography), cardio immunology, spectrophotometry and other methods for identifying and determining the exact amount of enzymes, hormones and other proteins;   | Basic foundations of each technique and their appropriateness, knowing the main biochemical tests for genetic disorders, how to interpret the results and their limitations;  |
| Pre-natal Diagnostics (PND)     | Follow the main methods used in obstetrics for diagnosis or in foetal monitoring (ultrasound scanning, amniocentesis, villus biopsy, cordocentesis and fetoscopy) and intentional termination (abortion); Learning to deal knowledgeably and sensitively in supporting couples having difficulty in conceiving;  | Acquisition of appropriate basic knowledge of embryology and of normal foetal development, including in general cases, knowing the more usual causes for multiple stillbirths and infertility, recognition of the more usual developmental abnormalities, their causes and prevention, understanding the meaning, advantages and limitations of obstetric results obtained from PND, understanding the importance of the interdisciplinary nature of PND.                                       |
| Paediatrics                     | Know how to observe the normal development of neonatals and children and recognise, appropriately and in good time, any deviations discovered; correctly conduct and write up physical examinations and obtain the relevant clinical and family history; establish a proper and effective relationship with young patients and their families;   | Knowledge of normal infant development in terms of weight and psycho-motor skills and recognition of any deviation: knowledge of the most common causes of slow development and delayed psychomotor development; know how to diagnose and deal with some of the more frequent genetic cases in paediatrics and know their causes and prevention;  |
| Internal medicine               | Correctly conduct and write up physical examinations and obtain the relevant clinical and family history of young or adult patients of various ages with various illnesses; establish a proper and effective relationship with adult patients and their families   | Know how to diagnose and treat some of the more frequent genetic cases in internal medicine or in the chosen adult medical specialty and know their causes and prevention;  |
| Optional – epidemiology         | Familiarisation with the main public health objectives regarding genetic disorders and with the main applications and methods relating to epidemiology and their practical results; participate in screening and recording human disorders;  | Acquisition of appropriate basic knowledge of human population structures and variations thereof; understand how such structures and variations can contribute to the disorder and its prevention; knowledge of how the network of primary health care services works and its relevance to planning treatment and prevention of disorders and the promotion of human health; knowledge of the main epidemiological methods and, in particular, those used in screening and recording epidemics; |
| Health statistics and ICT       | Become involved in the planning of and participate in studies concerning the collection, recording, updating and analysis of statistics relating to health; critical discussion of the methodology and conclusions of official scientific health data relating in particular to medical genetics;  | Knowledge of the process for collecting, recording and updating official statistics relating to health (vital statistics, demographic data etc); knowledge of the principal measurement methods and knowing how to interpret the results; knowing how to construct and test alternative hypotheses using the appropriate analysis procedures and interpret their results critically; understanding the main epidemiological problems, in particular those most relevant to medical genetics;    |

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| <p>Clinical genetics</p> | <p>(a) Observe patients and their families and diagnose or confirm diagnoses of genetic complaints, congenital abnormalities and dysmorphic syndromes;</p> <p>(b) Draw up and properly interpret pedigrees and identify family members at risk of genetic disorders;</p> <p>(c) Assess the risks of genetic disorder appearing or recurring, including manifestation and transmission of genetic predisposition;</p> <p>(d) Carry out genetic counselling sensitively and tactfully, using nondirective counselling techniques and without any preconception, communicate all the information necessary in each case, according to the psychological state, level of education, knowledge and moral beliefs of the patient, using thorough knowledge of the basic principles and of the appropriate techniques;</p> <p>(e) Know and present all the available reproductive techniques, including prenatal diagnosis and medically assisted reproduction, appropriate to each genetic situation and each patient;</p> | <p>(a) Proper grasp of variability of phenotypes under both normal conditions and as a result of disorder, incomplete penetrance and variations in how the gene is expressed, penetrance being dependent on age and genetic heterogeneity, and of the implications thereof for (clinical and laboratory) diagnosis and prognosis;</p> <p>(b) Know how to diagnose and know the means available for the treatment and prevention of the more frequent genetic disorders, including prenatal diagnostic techniques, screening programmes and early diagnosis, detection of carriers and pre-symptomatic diagnosis, knowing how to interpret the results and know their limitations;</p> <p>(c) Knowledge of how to build and maintain genetic disorders registers and know their importance for planning care and prevention, as well as their potential danger and the essential rules to ensure the strictest confidentiality.</p> <p>(d) Obtain solid knowledge of the essential concepts in the various areas of medical genetics which include, among others, molecular and biochemical genetics, immuno-genetics, cytogenetics and cellular biology, developmental genetics, embryology, teratology, dysmorphology and foetal diseases, psychosocial genetics and other aspects of the social sciences which inter-relate with genetics, such as cultural anthropology and historical genetics, behavioural and psychiatric disorder genetics, population genetics, genetic epidemiology and development;</p> <p>(e) Knowing how to consult genetic information in medical literature and in specialist databases, whenever necessary and in the best way possible;</p> <p>(f) Thorough knowledge of the psychological, legal and ethical aspects relating to the practice of medical genetics, including those relating to the keeping of genetic registers, PND, carrier diagnostics and the detection of mutations for late hereditary disorders which are incurable or of susceptibility genes;</p> <p>(g) Adequate knowledge of the international recommendations and national legislation regarding PND and abortion, carrying out genetic testing (diagnostic, predictive and prenatal tests) and the keeping of records and files with genetic information.</p> |
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until further notice.