



**UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES
EUROPEAN UNION OF MEDICAL SPECIALISTS**

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Description of Clinical Genetics as a medical specialty in EU Aims and objectives for specialist training

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Specialty Profile

Clinical Genetics describes the medical elements of Genetics Services provided to individuals and families (and sometimes populations). Other components include laboratory genetics (cytogenetics, molecular genetics, and biochemical genetics), genetic counselling and academic genetics. The core activities of a genetic service can be defined as 'integrated clinical and laboratory services, provided for those with/concerned about a disorder with a significant genetic component (both inherited and sporadic). Due to the sharing of genes among family members, the whole family, not only the individual, represents the core patient in clinical/medical genetics.

This document relates to medically qualified individuals intending to train in the specialty of Clinical/Medical Genetics. It recognises that there may be overlaps with training programmes for other genetic professionals (scientists and counsellors) and that there may be opportunities for joint training for periods of the course.

Entry criteria

This may vary from country to country but would generally include a specified period of general medical training to include adult +/- paediatric medicine prior to commencing specialty training in Clinical Genetics, "internship". Some countries may have a minimum period of training to be undertaken before specialisation.

Educational goals

Knowledge and Skills

- **Theoretical genetics/Basic Science** which may include
 - understanding cellular and molecular mechanisms that underpin human inheritance,
 - understanding patterns of inheritance and methods for risk assessment,
 - genetic epidemiology and biostatistics
- **Clinical/Medical knowledge and skills**
 - Pedigree construction.
 - Diagnosis, investigation and genetic management of individuals with both common and rare inherited/genetic diseases and their families.
 - Risk assessment and role in genetic testing.
 - Paediatric genetics including training in Dysmorphology (knowledge of common dysmorphic syndromes, their aetiology and the use of dysmorphology databases) and investigation of learning disability in children.
 - Adult genetics to include knowledge of late onset disorders and disorders with a significant genetic component presenting in adult life (including predictive testing).
 - Prenatal Genetics and knowledge about fetal development and teratogens
 - Population genetics, including genetic screening programmes
 - Special areas of genetics including
 - Inherited metabolic disorders
 - Neuro- and neuromuscular genetics
 - Cardiovascular genetics
 - Cancer genetics
 - Neurosensory genetics (visual and hearing conditions)
 - Pharmacogenetics
 - Other subspecialties of specific interest to the trainee
- **Genetic counselling and communication skills**
 - Training in genetic counselling for all types of genetic disease and situations encountered in clinical genetic practice. This includes counselling in relation to prenatal diagnosis and for late onset such as neurogenetic and cancer genetic disorders, including predictive testing. Where applicable, training in co-counselling with other professionals such as genetic counsellors.
 - Understanding ethical issues and importance of consent and confidentiality.
 - Development of good communication skills with patients, colleagues in genetic centres and other specialists and healthcare professionals, including understanding and handling of crisis reactions.

- **Laboratory skills**
 - Thorough knowledge of principles of laboratory techniques used in diagnostic testing
 - Interpretation of results from cytogenetic, molecular genetic and biochemical genetic analyses.
 - The time spent and the practical expertise gained in laboratory work may vary between countries, but sufficient to ensure highly specialised knowledge.

Other aspects of the Training Programme

- **Maintaining Good Medical Practice**
 - Develop a commitment to lifelong learning through continuing professional development and attend relevant courses and conferences.
 - Participate in Audit and Clinical Governance
 - Adhere to established consent and confidentiality procedures
 - Understand ethical and legal issues
- **IT skills**
 - Use of information technology including online resources and databases
- **Management training**
 - Knowledge about general healthcare policy, goals and priorities
 - Understanding the organisation of genetic services
 - Opportunities to participate in departmental activities related to organizational planning, financial management, and monitoring and maintaining quality standards
 - Development of multidisciplinary team working and leadership skills
- **Teaching**
 - Develop teaching skills by participating in the education and training of various categories of staff
 - Involvement with patient groups and patient education
- **Supplementary Education and Training**
 - Subspecialty training: some trainees will elect to develop expertise in a subspecialty area such as cancer genetics, dysmorphology or neurogenetics.

Quality Assurance

- Competency-based curricula should form the basis of a training programme.

- A written agreed curriculum for the training period should be set up as a contract between the trainee and the supervisor if not otherwise determined by national regulations.
- Trainees should maintain a Training Log including details of clinical and laboratory experience, educational activities, research and publications.
- A mechanism should be in place for continuous assessment of trainees against agreed quality standards. Some countries will have a nationally prescribed system for assessment and certification.
- Specialist examination may be compulsory in some countries

Research

- Medical genetics has a rapidly changing knowledge base and during specialty training the clinical/medical geneticist should be encouraged to participate in research. Some trainees will wish to take time out from the clinical training programme to undertake an intensive period of research leading to a higher academic degree. On completion of training some academic clinical/medical geneticists will continue to lead research programmes whilst many others will collaborate with laboratory based colleagues in the genetics team.

Time frame for specialist training

- The training period should minimum 4 years full time work; part time work would extend the training period.
- An educational training programme will be agreed for each trainee according to the specialty specific curriculum
- In the longer training period, up to one year could be in another speciality of importance for clinical/medical/medical genetics.
- The time spent in laboratory work may vary between countries according to national curricula.
- A period of research resulting in a PhD/other higher exam may, if appropriate, replace training for a variable period of time according to national guidelines. However, in absence of national guidelines, it is not recommended that this time period is longer than 1/3 of the total training period