



UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES EUROPEAN UNION OF MEDICAL SPECIALISTS

Association internationale sans but lucratif

International non-profit organisation

RUE DE L'INDUSTRIE, 24
BE- 1040 BRUSSELS

T +32 2 649 51 64
F +32 2 640 37 30

www.uems.eu

info@uems.eu

UEMS 2017/06A

Description of Clinical Genetics as a Medical Specialty in EU Aims and objectives for specialist training

Endorsed by: The UEMS SCG Board and SCG Section Meetings (21 May, 2016, Barcelona)

Adopted/approved by: The UEMS Council (27-28 April, 2017, Tel Aviv)

Specialty Profile

Clinical Genetics is the medical specialty concerned with the medical elements of Genetics Services provided to individuals and families (and sometimes populations) with, or at risk of, conditions which have, or may have, a genetic basis. This includes the provision of diagnostic and genetic counselling services, information about the condition and its implications, including management, prognosis, screening, prevention and reproductive options, and therapeutic possibilities. This is based on thorough clinical assessment, family (pedigree) medical information, conventional laboratory investigations and imaging, and specialized genetic tests and their interpretation. Other components of Genetics Services include laboratory genetics (cytogenetics, molecular genetics, biochemical genetics, and genomics), specialized 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, or 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 areas of overlap with training programmes for other genetic professionals (scientists and counsellors) and that there may be opportunities for joint training for periods of the course.

Due to the differences in European health care systems across member states, there may be different national emphases to the different elements of this training programme.

These are minimum criteria reflecting the differences in training in the different EU countries.

UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES EUROPEAN UNION OF MEDICAL SPECIALISTS

Association internationale sans but lucratif – International non-profit organisation

Entry criteria

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

Educational goals

Knowledge and Skills

Basic theoretical genetics / Basic science

- Cellular and molecular mechanisms that underpin human inheritance
- Chromosome structure and function, mitosis and meiosis, and the origin of aneuploidy and other imbalances
- The structure of DNA and RNA, replication, transcription and translation. Embryology and the genetic determinants of human development
- Genetic epidemiology and biostatistics
- Population genetics, the principles of screening, and basic mathematical genetics
- Risk assessment, including Bayes’ theorem
- Bioinformatics
- Epigenetics
- Pharmacogenetics / -genomics
- Principles of acquired genetic disorders
- History of Genetics

Clinical/Medical knowledge and specialist-level skills

- Common and unusual patterns of inheritance
- Taking a detailed medical and family history
- Able to perform genetic risk assessment, including the use of Bayes’ Theorem to incorporate conditional risk information
- Able to undertake risk assessment in relation to genetic testing and apply the principles of specificity and sensitivity
- Pedigree construction and interpretation.
- Diagnosis, investigation and genetic management of individuals with both common and rare inherited/genetic diseases and their families.
- Therapeutic aspects and emerging therapies of genetic diseases
- Paediatric genetics including training in Dysmorphology (knowledge of common dysmorphic syndromes, their aetiology and the use of dysmorphology databases), and investigation of learning and intellectual disability in children.

UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES EUROPEAN UNION OF MEDICAL SPECIALISTS

Association internationale sans but lucratif – International non-profit organisation

- Adult genetic disorders, including knowledge of late onset diseases and conditions with a significant genetic component presenting in adult life
- Prenatal Genetics, fetal dysmorphism, and knowledge of the effects of common teratogens on fetal development
- Genetic screening programmes and newborn bloodspot tests
Competent clinical examination of both paediatric and adult patients, especially in relation to dysmorphic signs and features, and neurological examination and interpretation
- Gene therapy, its current and future applications, and other strategies for the treatment of genetic disease.
- Common diseases with a genetic component
- Multifactorial/polygenic disorders
- Genetic disorders that constitute part of mainstream speciality
- Sub-specialty areas of Clinical Genetics, including:
 - Inherited metabolic disorders
 - Neurogenetics and neuromuscular genetics
 - Cardiovascular genetics
 - Cancer genetics
 - Reproductive genetics
 - Other subspecialties of specific interest to the trainee e.g. connective tissue, immunogenetics, etc.

Genetic counselling and communication skills

- Training in genetic counselling for all types of genetic disease and genetics-related situations encountered in Clinical Genetics practice. This includes pre- and post-testing counselling in relation to reproductive options, prenatal diagnosis, and for late onset such as neuro- and cancer genetic disorders, including predictive genetic testing. Where applicable, training in co-counselling with other professionals such as genetic counsellors and specialists in other fields of medicine
- Understanding, and handling, emotional reactions and personal and family crises in relation to the impact of genetic disease and the genetic diagnostic process.
- Understanding ethical, legal and social issues, and importance of consent and confidentiality
- Development of good communication skills with patients and families, colleagues in genetic centres and other specialists and healthcare professionals

Laboratory skills

- Thorough knowledge of principles of classic laboratory techniques used in genetic diagnostic testing
- Thorough knowledge of the new laboratory techniques used in genetic diagnostic testing, including SNP and CGH arrays, whole genome sequencing and exome sequencing

UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES EUROPEAN UNION OF MEDICAL SPECIALISTS

Association internationale sans but lucratif – International non-profit organisation

- Interpretation of results from cytogenetic, molecular genetic, biochemical genetic and genomic analyses (array, exome and whole genome analyses)
- The time spent and the practical expertise gained in laboratory work may vary between countries, but it should be sufficient to ensure highly specialized knowledge to make eligible the candidates have to have technical skills to make them eligible to prepare and sign diagnostic and laboratory reports. They have to be able to perform these tests on a daily base. They have to have the capability to supervise technical staff's work.
- Knowledge of preanalytical handling of samples and logistics
- Awareness of quality issues in genetic testing
- Knowledge of international nomenclature systems used in genetic reporting.

Other aspects of the Training Programme

Maintaining Good Medical Practice

- Understand and practice medical professionalism, honesty, integrity, an aspiration to excellence, fairness, and avoidance of discrimination
- Develop a commitment to lifelong learning through continuing professional development and attend relevant courses and conferences.
- Participate in Audit and Clinical Governance
- Adhere to accepted consent and confidentiality procedures
- Timely management of medical documentation and communication with patients families and professionals'

IT skills

- Use of information technology including online resources and databases related to human genetics

Ethics and law

- Understand ethical, legal and social issues in relation to genetic medicine
- Issues relating to patient confidentiality, consent and disclosure of results.

Biobanking

- Understand principles of biobanking
- Awareness of ELSI issues

Management training

- Knowledge of national laws relating to genetic services and practice, general healthcare policy, goals and priorities
- Understanding the organization and management of genetic services
- Opportunities to participate in departmental/service activities related to organizational planning, financial management, and monitoring and maintaining quality standards
- Development of multidisciplinary team working and leadership skills

UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES EUROPEAN UNION OF MEDICAL SPECIALISTS

Association internationale sans but lucratif – International non-profit organisation

Teaching

- Develop teaching skills by participating in the education and training of various categories of staff
- Involvement with patient groups, and patient/family education

Supplementary Education and Training

- Subspecialty training: Some trainees will elect to develop expertise in a subspecialty area such as cancer genetics, dysmorphology, neurogenetics, etc. This may also vary from country to country.
- Knowledge and understanding of the principles of evidence-based medicine
- Involvement and initiatives in courses, programmes and social issues related to rare diseases
- Knowledge of patient registries, patient support organisations

Quality assurance

- Knowledge, skills and attitudes should form the basis of the 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 Logbook including details of clinical and laboratory experience, all 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
- EU-Certification in Clinical Genetics should be encouraged

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 may wish to participate in scientific projects and 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 wider genetics team.
- Understand the principles of research methodology including clinical trials

UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES EUROPEAN UNION OF MEDICAL SPECIALISTS

Association internationale sans but lucratif – International non-profit organisation

Time frame for specialist training

- The training period should last a minimum of 4 years full time work, of which one additional year could be done in another specialty before, after, or as a part of the specialist training. 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 (5 years), up to one year could be in another speciality of importance for Clinical/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.