



European Society of Human Genetics

Ad hoc committee for the accreditation of clinical laboratory geneticists

Clinical Laboratory Genetics as an EU-recognized specialist profession **Aims and objectives for specialist training**

Speciality Profile

The speciality profession Clinical Laboratory Genetics comprises all elements of genetic laboratory services needed to support patient or family handling by clinical/medical genetic specialists (see UEMS 2009/15), genetic counsellors and a wide range of other medical disciplines such as paediatrics, obstetrics and clinical oncology. Clinical Laboratory Genetics is a laboratory speciality on an academic level that joins cytogenetics, oncogenetics, molecular genetics and metabolic/biochemical genetics into one skilled profession. The specialist has deep knowledge of laboratory services and results interpretation related to genetic disorders, both inherited and sporadic.

Target group

This document relates to individuals qualified in natural sciences (e.g. biology and related subjects) and/or medicine intending to become Clinical Laboratory Geneticist. Since there may be overlaps with training programs for other medical genetic professions (clinical/medical genetics for MD's and genetic counsellors/nurses), an opportunity for sharing some training modules is present.

Entry criteria

This may vary somewhat from country to country but would generally include a specified period of general natural sciences and/or medical training with a significant component of genetics to ensure basic biological and/or medical knowledge prior to commencing specialty training in Clinical Laboratory Genetics. In all countries a minimum entry level, like a related M.Sc. degree, should be obtained before entry into such a speciality program may be considered.

Exit criteria

Knowledge and skills obtained during the training program for Clinical Laboratory Geneticist should be the following:

- A. Relevant and sufficient scientific knowledge in biology and genetics:**
- Understanding cellular and molecular mechanisms that underpin human inheritance.
 - Basic knowledge in clinical terminology, epidemiology, statistics and cell biology.
 - Knowledge of patterns and modes of inheritance (Mendelian and non-Mendelian diseases, genomic disorders, epigenetics, genetic modifiers, multifactorial disorders, variable penetrance and expressivity), issues related

to reproductive choices and family studies, and specifically awareness of the appropriate methods for testing such problems and their limitations.

- Familiarity with basic as well as modern test systems to characterize the human genome at different levels of resolution, including a full understanding of the advantages, disadvantages and limits of each system.
- Understanding of DNA/cellular/genetic function to enable optimisation, validation and implementation of new test systems.
- Knowledge about how to organize and lead a laboratory.
- Basic knowledge on metabolic disorders.
- Knowledge on bioinformatics and biobanking issues.
- Familiarity with issues related to quality assurance (OECD recommendations, ECA guidelines, Eurogentest – EQA – recommendations, national guidelines), accreditation (ISO 15189).

B. Clinical/Medical knowledge and skills:

- Means of diagnosis, differential diagnosis, investigation and genetic testing for individuals with both common and rare inherited/genetic diseases and their families.
- Knowledge on mutagenesis and teratogenesis, e.g. possible effects of radiation, toxins, mutagens and prenatal infections.
- Knowledge of prenatal tests, postnatal and oncology tests including methods, risks and limitations.
- Knowledge of common international nomenclature systems (like ISCN, HGVS) and online resources (like OMIM, Orphanet and GeneReviews).
- Knowledge of clinical indications and the clinical utility and cost effectiveness of various testing procedures.
- Evaluation of family information, how to construct pedigrees and calculate inheritance risks.
- Issues related to reproductive choices and family studies, and specifically awareness of the appropriate methods for testing such problems and their limitations.
- Deeper knowledge and specialized training in elected main subjects of genetic laboratory diagnostics, e.g.
 - o Inherited cancer genetics
 - o Sporadic cancer genetics, including haematology
 - o Clinical Genetics
 - o Metabolic diseases
 - o Developmental disorders
 - o Cardiogenetics
 - o Neurogenetics
 - o Other subspecialties of specific interest to the trainee
- Knowledge on medical ethical issues in diagnostics and research (e.g. informed consent, privacy, autonomy)

C. Team and communication skills:

- Development of good communication skills with colleagues in genetic centres and other specialists and healthcare professionals
- Ability to issue reports of not only biological but also clinical validity, relevant for patient diagnostics and handling.
- Effective communication with laboratory staff for efficient service provision, including teamwork and leadership abilities.

- Understanding ethical issues and importance of consent and confidentiality.
- Where applicable, co-counselling training with other professionals such as specialist in clinical/medical genetics or genetic counsellors.

D. Laboratory skills:

- Thorough laboratory practice (including hygiene and safety) and knowledge on how to run and organize a laboratory (some countries may have a minimum period of training and/or case numbers to be studied in different special fields; the Dutch curriculum is recommended as orientation for national requirements).
- Knowledge on how to handle, identify, proceed, transport and archive samples and test materials, including knowledge on biobanking.
- Prioritise sample testing to reflect clinical urgency.
- In depth knowledge on interpretation of genetic laboratory findings, more specifically results from cytogenetic, molecular cytogenetic, oncogenetic, molecular genetic and biochemical genetic analyses.
- Practical experience and background knowledge of the (pre)analytical techniques and methods to be applied in genome diagnostics: cell culture, DNA isolation, karyotyping and specific staining techniques, FISH, PCR, copy number variations, gel electrophoresis, Sanger sequence analyse, new sequencing technologies, array technologies, Southern blotting technique, scanning analysis (HRMCA, heteroduplex analysis, DGGE, SSCP, PTT, etc.), fragment length analyses and RNA examination.
- Reporting: preparing reports of results
 - o can produce a complete and adequate description of the tests;
 - o can independently interpret the results of the test in the context of the diagnostic question and formulate a conclusion, if necessary with the help of additional techniques or after consulting the literature;
 - o knows the most important implications of the diagnoses;
 - o is aware of the possibilities and limitations of the applied techniques and of their causes;
 - o understands the positive or negative predictive value of a test;
 - o integrates multidisciplinary data in a good way;
 - o integrates the national and international developments in the molecular and cytogenetic areas in the diagnostics;
 - o is aware that with more difficult case histories, internal or external consultation can often be essential;
 - o produces comprehensible reports in compliance with internationally recognised standards.
- Awareness of new technologies:
 - o draw attention to potential new methods, techniques, developments in bioinformatics;
 - o imaging diagnostic possibilities for new methods;
 - o estimating efficacy, quality and cost of new methods;
 - o preparing implementation criteria for new techniques;
 - o validating new methods and techniques.
- Skills to develop, optimise, validate and introduce new investigations into diagnostic service
- Familiarity with issues related to quality assurance (OECD recommendations, ECA guidelines, Eurogentest – EQA – recommendations, and national guidelines), accreditation (ISO 15189) and clinical governance.

E. Knowledge of Good Medical Practice:

- Participate in audits and clinical quality procedures.
- Adhere to established consent and confidentiality procedures.
- Understand ethical and legal issues.
- Promote lifelong learning through continuing professional education and relevant course/conference attendance.

F. IT skills

- Acquire knowledge on how to exploit IT-technology for promoting good laboratory work and result interpretation, related to in-house software solutions, online resources and databases.
- Knowledge on data safety.

G. Management training

- Development of multidisciplinary team working and leadership skills.
- Understanding the organisation of health- and clinical genetic services.
- Opportunities to participate in departmental activities related to organizational planning, financial management, and monitoring and maintaining quality standards.
- Knowledge about general healthcare policy, goals and priorities.

H. Teaching

- Promote teaching skills by participating in the education and training of students and health care workers, if possible also patient groups.

Quality Assurance

- 1) 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 (see e.g. the Dutch curriculum regarding national requirements).
- 2) All trainees should maintain a Training Log, preferably in an electronic and transferable format, including details of clinical and laboratory experience, educational activities, research and publications.
- 3) 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.
- 4) Specialist examination may be compulsory in some countries

Research

Clinical Laboratory Genetics has a rapidly changing knowledge base and the trainees should be encouraged to participate in research.

Time frame for specialist training

- 1) The training period should be minimum 4 years full time work. Part time work would extend the training period

- 2) An educational training programme will be agreed for each trainee according to the specialty specific curriculum.
- 3) The time spent in laboratory work, number of issued reports and number of necessary sub-specialisations (module-based system) may vary between countries depending on national curricula.
- 4) 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 1/2 of the total training period.

At the end of this period, extended laboratory practise and training is usually needed before the individual is fully competent to independently run a laboratory service. It must be ensured that knowledge is up-to-date and that experience in case investigations and result interpretation is sufficiently comprehensive for giving independent clinical advice.