EBMG European Board of Medical Genetics

European registered Clinical Laboratory Geneticist (ErCLG)
Core curriculum

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Speciality Profile

The speciality profession European registered Clinical Laboratory Geneticist (ErCLG) comprises all elements of genetic and genomic laboratory services related to the provision of medical genetics services to patients or families 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, cancer genetics, molecular genetics/ genomics and metabolic/biochemical genetics into one skilled profession. The specialist has deep knowledge of and core-competences in laboratory services, procedures, results interpretation, and reporting related to a) genetic disorders, both inherited and acquired, and (b) health issues related with genetic factors.

Target group

This document relates to individuals qualified in biosciences (e.g. biology and related subjects) and/or medicine intending to become ErCLG. 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, such as 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 ErCLG should include the following:

A. Relevant and sufficient scientific knowledge in biology and genetics:

- Understanding cellular and molecular mechanisms that underpin human physiology and inheritance.
- Familiarity with multi-omics approaches (transcriptomics, proteomics, metabolomics) and their integration with genomic data
- Basic knowledge in clinical terminology, epidemiology, statistics, molecular 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).
- Basic knowledge of differential transcript usage, alternative splicing, and isoform-specific effects and its impact on gene function, tissue specificity, and phenotypic variation.
- Basic knowledge on chromosome biology and nomenclature (ISCN).
- Basic knowledge of variant nomenclature using the HGVS recommendations and variant interpretation and classification (ACMG Guidelines).
- Familiarity with basic as well as current 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 central dogma of molecular biology in the DNA/RNA/protein level and its cellular/genetic function to enable optimisation, validation and implementation of new test systems.
- Basic knowledge of metabolic disorders.
- Knowledge of bioinformatics and biobanking issues.

B. Clinical/Medical knowledge and skills:
- Basic knowledge of human anatomy and medical terms.
- Means of prognosis, diagnosis, differential diagnosis, investigation and genetic and genomic testing for individuals, families or populations with a) both common and rare inherited/genetic (acquired) diseases, b) disease related genetic factors.
- Knowledge of genotype-phenotype relationships for certain genetic disorders and health issues related with genetic factors.
- Knowledge of 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, ACMG Guidelines) and online resources (like OMIM, Orphanet, GeneReviews, DECIPHER, ClinGen and others).
- Knowledge of clinical indications and the clinical utility, limitations 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.
- Basics of polygenic risk calculation and interpretation.
- Deeper knowledge and specialized training in elected main subjects of genetic and genomic laboratory diagnostics, e.g.
    - Inherited cancer genetics

    - Sporadic cancer genetics, including haematology
    - Clinical genetics
    - Prenatal testing and diagnostics
    - Inborn errors of metabolism
    - Reproductive genetics
    - Developmental disorders
    - Cardiovascular genetics
    - Neurogenetics and neuromuscular genetics
    - Gene therapy and genome editing
    - Pharmacogenomics
    - Other subspecialties of specific interest to the trainee
- Knowledge of medical/genetic testing ethical issues, and national and EU jurisprudence in diagnostics and research (e.g. informed consent, privacy, autonomy, incidental and secondary findings, general data protection regulation (GDPR), etc.).
- Knowledge of the principles and practice of pre-test and post-test genetic counselling, including informed consent, communication of benefits, limitations and possible outcomes of testing, and implications for the individual and family.
- Knowledge of patient opt-in/opt-out choices regarding receipt of genetic results, especially secondary or incidental findings, and their implications for informed consent and counselling.

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 provision of medical care.
- 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:
- Knowledge of how to organize and lead a laboratory.
- Familiarity with quality assurance requirements (OECD recommendations, ECA guidelines, Eurogentest – EQA – recommendations, national guidelines), accreditation (ISO 15189).
- Thorough laboratory practice (including hygiene and safety) and knowledge of 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 of how to handle, identify, proceed, transport and archive samples and test materials, including knowledge of biobanking.
- Prioritise sample testing to reflect clinical urgency.
- In depth knowledge of interpretation of genetic and genomic laboratory findings, more specifically results from cytogenetic, molecular cytogenetic, oncogenetic, molecular and biochemical genetic analyses. Correlation with phenotypic data.
- Practical experience and background knowledge of the pre-analytical and analytical techniques and methods to be applied in genome diagnostics: cell

culture, DNA isolation, karyotyping and specific staining techniques, FISH, PCR/qPCR/digitalPCR incl. MLPA, copy number variations, gel electrophoresis, sequencing technologies, including Sanger sequencing, next generation sequencing, and long read sequencing, array technologies (aCGH/CMA/seq-CNV), optical genome mapping (OGM), RNA-based analyses, methylation and epigenetic testing methods, and repeat expansion testing.

- Reporting: preparing reports of results
    - can produce a complete and adequate description of the tests;
    - 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/databases/guidelines;
    - knows the most important implications of the diagnoses;
    - is aware of the possibilities and limitations of the applied techniques and of their causes;
    - understands the positive or negative predictive value of a test;
    - integrates multidisciplinary data in an appropriate way;
    - integrates the national and international developments in the molecular and cytogenetic areas in the diagnostics;
    - is aware that with more difficult case histories, internal or external consultation can often be essential;
    - produces comprehensible reports in compliance with internationally recognised standards;
    - is able to incorporate incidental and secondary findings, when appropriate, helpful for interpretation and necessary.
- Awareness of new technologies:
    - be aware of emerging new methods, techniques and developments e.g. in bioinformatics;
    - appreciate diagnostic possibilities for new methods;
    - estimating efficacy, quality and cost of new methods;
    - preparing implementation criteria for new techniques;
    - validating new methods and techniques e.g. RNA sequencing and functional genomics as complementary tools to resolve VUS.
- Skills to develop, optimise, validate and introduce new investigations into diagnostic service

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 of how to exploit IT-technology for promoting good laboratory work and result interpretation, related to in-house software solutions, online resources and databases.
- Knowledge of data safety, including general data protection regulation (GDPR).

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 of Training

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.

National training programs

National training programs should acknowledge the holders of the title “European registered Clinical Laboratory Geneticist (ErCLG)”. It is suggested that national professional organisations develop regulations for ErCLGs who move from the country where they obtained the registration as an ErCLG, to a new host country. The host country presupposes, for example, award the ErCLG a national CLG title after the ErCLG has worked there for an appropriate period of time, such as 3 years and fulfilling the mandatory national requirements (e.g. compliance with the knowledge of the state language).

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https://www.ebmg.eu/clg.0.html

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