**Cyprus registered Clinical Laboratory Geneticist (CyrCLG) Core curriculum**

(January 2016; paper issued by the Cyprus Society of Human Genetics Ad hoc committee for the accreditation of clinical laboratory geneticists, Cyprus Board of Medical Genetics)

**Speciality Profile**

The speciality profession Cyprus registered Clinical Laboratory Geneticist (CyrCLG) comprises all elements of genetic laboratory services related to the provision of medical genetics services to patients or families by clinical/medical genetic specialists. 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, procedures and results interpretation related to a) genetic disorders, both inherited and sporadic, 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 CyrCLG. Some training modules are suitable for other genetic professions such as clinical/medical genetics for MD’s and genetic counsellors/nurses.

**Entry criteria**

Entry criteria include both professional and academic qualifications.

**A.** Academic: Minimum a related MSc degree

**B.** Professional: At least two years of general natural sciences and/or medical training.

**Exit criteria**

Knowledge and skills obtained during the training program for CyrCLG 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.

- 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).

- 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 of how to organize and lead a laboratory.

- Basic knowledge of metabolic disorders.

- Knowledge of 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 prognosis, diagnosis, differential diagnosis, investigation and genetic testing for individuals, families or populations with a) both common and rare inherited/genetic 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)

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, as defined in the sub-specialty curriculum.

* Biochemical Genetics
* Cytogenetics
* Molecular Genetics
* Oncogenetics

- Knowledge of 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 health and safety) and knowledge of how to run and organize a laboratory (a minimum of 5 years training and a defined number of cases according to the specialty specific curriculum).

- 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 laboratory findings, more specifically results from cytogenetic, molecular cytogenetic, oncogenetic, molecular genetic 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 according to the specialty specific curriculum

- 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;
* 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 specific specialty 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.

- Awareness of new technologies:

* be aware of potential new methods, techniques, developments 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.

- 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.

**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 in an accredited laboratory should be set up as a contract between the trainee and the supervisor.

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) CSHG has documents for continuous assessment of trainees and trainers against agreed quality standards.

4) Specialist examination may be used as an additional evaluation tool.

**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 5 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 number of issued reports may differ between the sub-specialisations (module-based system) and is specified in the respective curricula.

4) A period of research resulting in a PhD/other higher exam may, if appropriate, replace training for a variable period of time. However, this time period may not be longer than 2 years.

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.

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