TRAINING PROGRAMME
in the field of
MEDICAL GENETICS

1. Purpose of specialist training

The purpose of specialist training is the acquisition of comprehensive qualifications in the field of medical genetics which allow those who have completed the training to work independently at workplaces which specialise in medical genetics and to do so in close cooperation with cytogenetics and molecular genetics laboratories and with workplaces which specialise in biochemical genetics.

2. Minimal requirements for specialist training

Entry to specialist training in the field of medical genetics is conditional on obtaining professional qualifications to practice medicine by completing a course of full-time study which lasts for at least six years and covers theoretical and practical instruction as part of an accredited master's degree programme in general medicine at a medical faculty.

Specialist training is undergone while practising medicine and takes the form of round-the-clock training to an extent corresponding to the weekly working hours stipulated in § 83a of the Labour Code.

Acquiring a specialisation in the field of medical genetics is conditional on being assigned to the field and completing practical training within a minimum total period of four to five years in accordance with the duration of the common core curriculum for the relevant fields.

2.1 Compulsory core curriculum – *minimum of 24 months* of practical training

Completion of introductory compulsory practical training as part of the common core curriculum for internal medicine (paediatrics), gynaecology and obstetrics, and where necessary of the common curriculum for other fields. The requirements for the specialist practical training, compulsory training activities, knowledge and skills which are essential to completion of the common core curriculum are laid down in the relevant training programme.

2.2 Specialist training – *minimum of 24 months*

a) compulsory practical training in the field

18 months minimum: practical training in the field of genetic outpatient care in selected accredited workplaces specialising in the field of clinical genetics, including oncogenetics

of which: *12 months* in an accredited workplace,

1 week in a workplace specialising in prenatal diagnosis,

b) compulsory supplementary practical training

2.5 months: cytogenetics laboratory for classical and molecular cytogenetic methods,

3 months: selected molecular genetics laboratory,

1 week: selected workplace specialising in the field of biochemical genetics,

1 week: selected workplace specialising in assisted reproduction or reproductive genetics,

c) participation in training activities

- course in developments in medical genetics – *2 days,*
- specialisation internship – *2 × 1 week,*
- course in the psychology of genetic counselling – *1 day,*
- compulsory course in medical first aid (*3 days*) and a seminar on the foundations of health legislation (*1 day*), if not completed during the previous training programme,
- recommended participation in at least one international course in genetics,
- participation in specialist seminars and conferences organised by the Society for Medical Genetics of the Jan Evangelista Purkyně Czech Medical Association and in training activities organised by the IPVZ (Institute for Postgraduate Training in Health) and other organisations (e.g. ČLK (Czech Medical Chamber)).

3. Scope of required theoretical knowledge and practical skills

3.1 Scope of required theoretical knowledge

- Fundamental principles and concepts of general and clinical medicine.
- Types of heredity, mitochondrial heredity, genotype, phenotype, imprinting, chromosomal basis of heredity, autosomes, gonosomes, chromosomal mutation in clinical genetics, microdeletion.
- Classification of genetic diseases. Genetics of complex diseases.
- Fundamental principles and concepts of molecular genetics. Central tenets of molecular biology, structure of DNA, replication, transcription, translation, reverse transcription, genetic code, the gene, mutation and polymorphism — implications, repair mechanisms, genome, gene mapping.
- Human genome project — opportunities and constraints for clinical genetics, transcriptome, proteome.
- Mutagenesis, teratogenesis.
- Methods of molecular genetics, classical and molecular cytogenetics.
- DNA extraction, blotting methods, PCR and its modification, direct and indirect DNA diagnosis, DNA sequencing, interpretation of results.
- Classical cytogenetic methods, chromosome classification, types of banding. Principles of molecular cytogenetics, types of methods, CGH.
- Foundations of population genetics, Hardy Weinberg law, frequency of the most common defects and genetic diseases in the Czech Republic.
- Foundations of biochemical genetics. Taxonomy of hereditary metabolic disorders (HMDs), frequency of HMDs among the Czech population, types of heredity, population screening and selective screening for HMDs, basic clinical and diagnostic characteristics of lysosomal, peroxisomal and mitochondrial disorders and of disorders of the amino acid and saccharide metabolism, differential diagnosis and treatment of peracute or intermittent HMDs, determination of heterozygotes and prenatal diagnosis of HMDs.
- Foundations of the genetics of the immune system.
- Foundations of oncogenetics, molecular genetics of cancerogenesis.
- Genetic counselling. Genealogy, assessment of risk recurrence, familial marriages, dysfertility. Specialist genetic counselling in the field of oncogenetics, reproductive

genetics and other areas. Preventive methods in clinical genetics, preimplantation diagnosis and prenatal diagnosis, biochemical and ultrasound screening. Registration of congenital developmental defects and of the results of prenatal diagnosis, implications.

- *In vitro* fertilisation methods.
- Treatment of genetic diseases. Gene and cell therapy.
- Ethical problems in clinical genetics. Ethics of genetic counselling, informed consent, ethics of predictive testing, ethics of prenatal diagnosis, termination of pregnancy, ethics of genetic testing.
- Genetics and society, enshrining genetic procedures in legislation.

Requirements regarding theoretical knowledge of related and neighbouring fields

- Foundations of medical psychology.

3.2 Scope of required practical knowledge

- Foundations of the clinical examination of disabled probands.
- Genealogical analysis.
- Analysis of classical and molecular cytogenetic characterisation of chromosomes. Microscopic analysis of chromosome orientation.
- Analysis of basic methods of direct/indirect DNA diagnosis, interpretation of results.

4. General requirements

Knowledge of the basic legal provisions currently in place regarding health care, of the health care system and of documentary material. The provision of health care using ionising radiation equipment requires completion of a certified course in radiation protection.

5. Assessment of specialist training

a) Continuous assessment by a supervisor – the supervisor assesses the intern during the internship on the basis of a schedule of specialist training and keeps records on the completion of practical training (of specific activities in the workplace).

b) Requirements for sitting the postgraduate examination

- completion of the common core curriculum and its assessment in the specialisation certificate, including successful completion of the written test,
- completion of compulsory training measures,
- essay on a subject in the field of clinical genetics.

c) The postgraduate examination

- practical part - proband case study from a family – genealogical analysis, diagnosis, genetic prognosis, prevention; analysis of classical and molecular cytogenetic characterisation of chromosomes; analysis of a specific DNA diagnosis, interpretation of the results,
- theoretical part - 3 technical questions.

6. Nature of the activities for which those who have completed specialist training are qualified

Those who have completed specialist training in medical genetics are able and authorised to provide individual genetic counselling, perform genetic examinations in a

consultative capacity, assess the results of prenatal screening tests, participate in prenatal diagnosis and indicate specialist genetic and clinical examinations and accessible preventive and therapeutic measures. They help to train other specialists in the field of medical genetics. Acquiring a specialisation in medical genetics is the minimum requirement for performing management functions in workplaces which specialise in medical genetics.