

CLINICAL GENETICS

1. GENERAL ASPECTS OF THE SPECIALISATION

1.1 Purpose of the specialisation

The specialisation is an education and learning process in which trainee specialists (residents) acquire theoretical and practical knowledge in the area of clinical genetics and genetic counselling so that they are capable of treating, on their own, most patients with genetic diseases. Genetic treatment is taken to mean clinical, cytogenetic and molecular-genetic diagnosis, genetic counselling, prevention and therapy.

As part of the specialisation, trainees may also follow a dedicated specialisation in the narrower field of clinical genetics in a specific field of medicine, such as gynaecology, paediatrics, neurology, dermatology, ophthalmology, oncology, etc. The dedicated specialisation also qualifies the trainee specialist for specific work in these clinical areas.

The dedicated specialisation in clinical genetics, which is presented below, is designed for graduates from the faculty of medicine and is one of the three planned dedicated specialisations within the specialisation of clinical genetics. The laboratory dedicated specialisations, for which the syllabus is currently being drawn up, are clinical cytogenetics and clinical molecular genetics.

1.2 Duration and structure of the specialisation

The specialisation in medical genetics lasts five years and comprises the following:

- initial training ('common core' of training), which lasts 2.5 years
- follow-on training, which lasts 2 years and
- an option, which lasts six months.

The initial training is the same for all trainee specialists in clinical genetics, whereas the follow-on training and the option are specific to each programme of narrower specialisation.

1.3 Completion of specialisation

The main mentor ascertains that the specialisation has been completed after checking its sufficient duration, the fulfilment of the stipulated conditions regarding knowledge acquired, the number and quality of procedures and the successful completion of the stipulated tests. The specialisation is concluded by a specialisation exam.

2. VERIFICATION OF KNOWLEDGE

Each trainee specialist has his own training booklet and a separate log in which he enters all the procedures carried out, the times he acted as first assistant and his contributions in the fields of expertise, education and research.

2.1 Continuous assessment of knowledge

In order to guarantee that the specialisation is of adequate quality, the knowledge and skills acquired by the trainee specialist are checked by permanent supervision and occasional assessment through tests.

The permanent direct or indirect supervision of the acquisition of knowledge and skills is the task of mentors. It is carried out regularly and on a daily basis.

The knowledge of trainee specialists is checked through tests after the completion of training in a given unit of study. Tests may be written or oral. Knowledge is checked by presenting patients to trainees and asking trainees to prepare seminars, give a review of the literature, write articles and take part in research.

At least once a year trainee specialists must publicly demonstrate the knowledge they have acquired, and do so in the manner determined on each occasion by the direct or main mentor:

- present an analysis of a group of patients or an interesting individual clinical case at a meeting of a group of experts in the training institution or elsewhere;
- prepare and manage a clinical or clinical pathology conference on a topic in the area of specialisation;
- publish an article on a topic covered by the programme of specialisation in a reviewed domestic or foreign medical journal.

In order to continue the specialisation, trainees must pass the tests and be given a positive annual assessment by their main mentor.

2.2 Specialisation exam

The exam tests the theoretical and practical knowledge acquired by the trainee specialist. The theoretical part comprises a written test and an oral test. The tests cover knowledge in the areas of clinical genetics, medical cytogenetics and medical molecular genetics and, where the specialisation involves a dedicated specialisation, the field of expertise concerned. The test of practical knowledge covers clinical genetics and, where the specialisation involves a dedicated specialisation, the field of expertise concerned.

3. SPECIALISATION PROGRAMME

The specialisation provides trainees with knowledge and skills in the basics of human genetics, clinical medicine, genetic counselling, laboratory medical genetics (cytogenetics, molecular genetics and biochemistry) and the basics of research work.

The planned training method and a link to the minimum number of genetic counselling sessions and laboratory tests (point 4), where appropriate, are set out under the title of each study topic.

Training through study workshops, courses and postgraduate lectures will take place outside regular working time.

3.1 Initial programme ('common core')

Duration: 2.5 years (27.5 working months, 2.5 months of leave)

The minimum duration of training in laboratory medical genetics is six months.

Content of the programme

a. Basics of human genetics

History of human genetics

Human genetics up to the 18th century, the contribution of Galton, Mendel and Garrod, the importance of the discovery of blood groups, the HLA system, population genetics

(Haldane, Fisher), the development of cytogenetics, molecular genetics, eugenics and human genetics policy, the history of human genetics in Slovenia

Training method: independent study, seminar work

Methods of inheritance and exceptions

Basics and particularities of autosomal dominant inheritance, autosomal recessive inheritance, X-chromosome linked dominant inheritance, X-chromosome linked recessive inheritance, mitochondrial inheritance, the mechanisms involved in these forms of inheritance, phenomena of genetic expressivity, penetrance, heterogeneity, phenocopies, pleiotropy, anticipation, genomic imprinting

Training method: independent study, study workshops, seminar work

Human molecular genetics and cell biology

Structure of the genome and the gene, gene expression, biological mechanisms of proliferation, differentiation, methods of study thereof

Training method: independent study, lectures as part of university post-graduate studies in biomedicine

Developmental biology and embryology

Basics of normal and pathological embryology, embryological mechanisms, pathogenetic mechanisms (malformations, deformations, disruptions, dysplasia), clinical classification of development abnormalities (system handicap, associations, sequences, syndromes)

Training method: study workshops, independent study, seminar work

Mathematical and population genetics

Basics of probability calculation, Bayesian calculation, Hardy-Weinberg law, importance of genetic polymorphisms, influence of new mutations, natural selection, migrations, random fluctuations, consanguinity and genetic drift in the frequency of genes and alleles

Training method: independent study, seminar work

Carcinogenesis

Cell cycle, apoptosis, tumour suppressor genes, oncogenes, mechanisms of carcinogenesis, genetic predisposition for cancerous diseases, frequency of cancerous genetic diseases, genetic diagnosis of cancerous diseases, importance of genetic markers for prognosis of cancerous diseases

Training method: independent study, seminar work

b. Basics of clinical medicine

Examination of a foetus, child and adult

General clinical examination and dedicated dysmorphological examination (anatomical regions, organ systems), anthropological measurements, medical photography

Training method: practical work in genetic outpatients department, period of work in paediatrics (examination of a child) and pathology (foetal autopsy)

Minimum coverage: 4.1-4.7

Diagnostic approaches and treatment of main disease conditions in pregnancy, childhood and adulthood

Clinical characteristics, differential diagnosis, methods of diagnosis and treatment/prevention of the most frequent genetic diseases in pregnancy, the neonatal period, childhood and adulthood

Training method: practical work in genetic outpatients department, training in clinics and departments that clinically treat patients with genetic diseases (neurology, paediatrics, gynaecology, dermatology, ophthalmology), regular meetings in training institutions with a presentation of cases

Minimum coverage: 4.1-4.7

Basics of evidence-based medicine

Use of the concepts of evidence-based medicine in access to medical information (use of MEDLINE, Internet), assessment of the relevance of published information especially in the areas of diagnostic tests, interventions and prognosis (assessment of meta-analyses, the recommendations in the literature and economic efficiency) and in medical decision-making techniques.

Training method: study workshop

c. Basics of clinical genetics

Production and analysis of genealogical charts

The information that genealogical charts must contain, the use of modern nomenclature for the compilation of genealogical charts, the interpretation of genealogical charts as regards the possible types of inheritance

Training method: practical work in genetic outpatients department

Minimum coverage: 4.1-4.7

Clinical treatment of development abnormalities

The frequency of the individual groups of development abnormalities, basics and particularities of analysis (medical history, dysmorphological examination, measurement, photography, laboratory tests, imaging), summaries of data (identification of key symptoms and signs, identification of a pattern, comparison with known cases, use of computer dysmorphology databases), possibilities of treatment/rehabilitation, genetic counselling, continuous monitoring of patients (medical surveillance and prevention of complications, modification of diagnosis)

Training method: practical work in a dysmorphology unit, clinical dysmorphology meetings, regular meetings at training institution with a presentation of cases, study workshop

Minimum coverage: 4.3

Clinical teratology

Frequency of teratogenic factors, categories and characteristics, factors that influence the expression of teratogenic potential, place of action, time of action of teratogenic factor, methods for identification of teratogens, use of computer databases in the area of teratology to assess risk and provide counselling

Training method: practical work in a teratology unit, study workshop, regular meetings at establishments of education with a presentation of cases

Minimum coverage: 4.3

Risk assessment for chromosomal, genic and multifactorial genetic diseases

Use of probability calculation (Bayesian calculation) to assess the risk of monogenic diseases, multifactorial diseases and diseases caused by chromosomal abnormalities, use of computer programs to calculate risk, use of empirical data

Training method: practical work in genetic outpatients department, regular meetings at training institutions with a presentation of cases

Minimum coverage: 4.1-4.7

Prevention of genetic diseases

Frequency of various categories of genetic diseases, importance of genetic counselling, methods of prenatal diagnosis (chorionic villus biopsy, amniocentesis, cordocentesis, placentocentesis, pre-implantation prenatal diagnosis, foetoscopy), screening tests (routine ultrasound, measurement of nuchal translucency, 3 hormone test), their importance for preventing chromosomal and gene abnormalities, primary prevention of complex genetic diseases

Training method: practical work in genetic outpatients department, regular meetings at training institutions with a presentation of cases, period of work in outpatients department for ultrasound diagnosis

Minimum coverage: 4.1-4.8

Treatment of genetic diseases

Symptomatic treatment, gene therapy, medical rehabilitation, self-help organisations

Training method: practical work in genetic outpatients department, seminar work

Minimum coverage: 4.1-4.7

Basic communications, application in genetic counselling

Basic principles of relationships in communication, the process and techniques of communication, identification and mastery of the personality of the discussion partner, interview technique, neuro-linguistic programming

Training method: course, practical work in genetic outpatients department

Minimum coverage: 4.1-4.8

Psychological bases of genetic counselling

Psychological aspects of genetic counselling, psychology of women, psychology of pregnancy, psychology of the family (family dynamics, therapy), mourning process, loss, influence of a handicapped child on the family, psychological aspects of infertility, adoption, use of psychodynamic theory of development, defence mechanisms

Training method: course, practical work in genetic outpatients department

Minimum coverage: 4.1-4.8

Population aspects of clinical genetics; screening tests, genetic registers

The importance of genetic factors in multifactorial diseases, importance of genetic variability in predictive and preventive medicine, characteristics of screening tests for genetic diseases, importance of genetic registers in preventing genetic diseases and ensuring the quality of medical care

Training method: course, practical work in genetic outpatients department, seminar work
Minimum coverage: 4.8

Research work

Independent research work in a research team, preparation of at least one scientific/research contribution

Training method: each candidate is given a topic of research or expertise at training institutions as part of the specialisation

d. Laboratory medical genetics

Basics of cytogenetic methods and FISH

Frequency of chromosomal abnormalities in various periods of life (prenatal, neonatal, paediatric, adult), structure and function of chromosomes, categories of chromosomal abnormalities, methods of analysis of normal and pathological chromosome structures, organisation of cytogenetics laboratory, genetic counselling in the case of chromosomal abnormalities

Training method: practical training in study laboratories, practical work in genetic outpatients department

Minimum coverage: 4.9

Basics of molecular genetic methods

Frequency of gene abnormalities in various periods of life (prenatal, neonatal, paediatric, adult), categories of mutations, molecular genetic methods, approaches to molecular genetic diagnosis, organisation of molecular genetics laboratory, genetic counselling after molecular genetics tests

Training method: practical training in study laboratories, practical work in genetic outpatients department

Minimum coverage: 4.10

Expected skill outcome

Independent genetic treatment of patients and families with most frequent genetic diseases under supervision of mentor.

3.2 Follow-on training programme

Duration: 2 years (22 working months, 2 months of leave)

Content of the programme

The follow-on training programme trains candidates to be 'general clinical geneticists', who are capable of providing genetic treatment to patients and families with genetic diseases that receive primary treatment in various branches of medicine, or to be clinical geneticists dedicated to the treatment of genetic diseases in a particular branch of medicine, e.g. paediatrics, gynaecology, neurology.

Trainee clinical geneticists who follow a dedicated specialisation gain more in-depth theoretical and practical knowledge in their chosen field of medicine in keeping with the programme of the specialisation for that field. They gain a more detailed insight into dedicated medical history in that field and dedicated examination, clinical diagnosis, in-depth clinical differential diagnosis, material collection methods, dedicated diagnosis

methods (e.g. imaging, electrophysiological), dedicated laboratory diagnosis (biochemical, enzymic, patho-histological, cell, molecular biological and molecular genetic), the construction of a composite detailed diagnosis and its likelihood, the provision of a prognosis and its likelihood, the choice of treatment (including prenatal treatment), the implementation, monitoring and assessment of the success of the treatment, rehabilitation, integrated patient management, documentation of the entire process and cooperation with various subspecialist teams.

For general clinical geneticists, the programme covers the objectives described above, but they are studied in less depth due to the time constraints imposed by training in several areas of expertise.

The recommended areas of expertise and the minimum duration of training:

Internal medicine	4 months
Paediatrics	4 months
Gynaecology	1 month
Neurology	1 month
Dermatology	1 month
Ophthalmology	1 month

Method of training: practical work in appropriate clinical areas depending on the chosen particular area of study, regular clinical meetings and talks with a presentation of cases, study workshops, symposiums, congresses

Minimum coverage: 4.11

Expected skill outcome

Independent genetic treatment of patients and families with genetic diseases under indirect supervision of the mentor.

3.3 Option

Duration: 6 months (5.5 working months, 0.5 months of leave)

Content of the programme

In the option, trainee specialists are trained in a narrower field of expertise, if at all possible in a genetic centre outside Slovenia. Alternatively, trainees can devote themselves to research work in the area of medical genetics and submit a report on the work done.

Expected skill outcome

Independent, fully trained expert in the field of clinical genetics, who is awarded the title of specialist after he has passed the exam.

4. MINIMUM NUMBER OF GENETIC COUNSELLING SESSIONS AND LABORATORY TESTS

The table shows the minimum number of genetic counselling sessions and laboratory tests in which the trainee specialist played an important role in treatment, as shown by the trainee specialist's training record.

GENETIC COUNSELLING	Number of sessions
<i>4.1 Chromosomal abnormalities</i>	
4.1.1 Down's syndrome	10
4.1.2 Others (at least 4 structural abnormalities)	30
<i>4.2 Genetic abnormalities</i>	
4.2.1 Cystic fibrosis	2
4.2.2 Haemophilia	2
4.2.3 Duchenne muscular dystrophy/Becker muscular dystrophy	5
4.2.4 Huntington's disease (at least 2 pre-symptomatic cases)	5
4.2.5 Metabolic diseases	2
4.2.6 Others (at least 10 pre-natal diagnoses)	50
<i>4.3 Pre-natal diagnosis of abnormalities</i>	
4.3.1 Chromosomal abnormalities	10
4.3.2 Genetic abnormalities	10
4.3.3 Pre-implantation genetic diagnosis	2
<i>4.4 Development abnormalities (dysmorphic syndromes)</i>	30
<i>4.5 Oncogenetics</i>	10
<i>4.6 Mental retardation</i>	10
<i>4.7 Mitochondrial diseases</i>	2
<i>4.8 Male/female infertility</i> (including spontaneous pregnancy loss)	10
<i>4.9 Screening tests for genetic diseases</i>	20
<i>4.10 Teratology</i>	10

LABORATORY TESTS	Number of tests
<i>4.11 Cytogenetic</i>	
Normal karyotype (blood, amniocytes, fibro-blasts)	10
High-resolution karyotype (blood)	5
Fluorescent in situ hybridization	5
<i>4.12. Molecular-genetic</i>	
PCR analysis (including direct and indirect molecular genetic analysis)	30
Southern blot analysis	5
MLPA	10

4.13. The number of treatments is increased in relation to the training programme as part of the follow-on training in the specialisation programme. The programme of the chosen course of study is drawn up by the clinical mentor in agreement with the trainee specialist's main mentor.

5. FORMS OF TRAINING:

- practical clinical and laboratory work,
- discussions with mentors,
- active participation in departmental meetings with handling of current clinical cases,
- seminars,

- participation in study workshops and recommended national and international meetings of experts, involvement in research.

Medical Chamber of Slovenia

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