

## SPECIALTY TRAINING IN CLINICAL GENETICS - SUMMARY

### 1. Objective:

This specialty training is aimed at giving doctors specific qualifications in the field of clinical genetics to enable them to treat patients with genetic diseases and their families in the light of current science on the subject, with particular emphasis on understanding the pathomechanisms of such diseases and their diagnosis and treatment. Clinical geneticists must also be able to carry out screening for the early identification of individuals and families with a high risk of contracting common diseases with a major social impact (malformations, familial cancers, certain metabolic diseases, etc.).

### 2. Knowledge required:

After completing the specialist training, a doctor should be able to demonstrate a knowledge of the following:

- a. the basics of medical genetics: molecular inheritance, cytogenetic inheritance, monogenic causes of inherited diseases, deviations from traditional Mendelian genetics, multigenic diseases, genetic risk, genetic diagnosis and counselling;
- b. cytogenetics;
- c. molecular genetics;
- d. clinical genetics in different medical specialties:
  - i. genetics in paediatrics: basics of paediatrics, genetic diseases in babies and infants, childhood genetic diseases;
  - ii. gynaecology and obstetrics; sex determination and differentiation, reproductive failure, diagnosis of the genetic burden;
  - iii. neurogenetics;
  - iv. oncogenetics;
  - v. genetic diseases of other organs and systems;
  - vi. pharmacogenetics.

### 3. Skills required:

After completing the speciality training, a doctor should be able to demonstrate the following skills:

- a. ability to construct and analyse lineages, knowledge of the principles and methods for obtaining diagnostic information;
- b. ability to plan and coordinate diagnostic tests in the light of the results of lineage analysis and consultation of specialists in different fields;
- c. ability to assess dysmorphic features and knowledge of the other principles of phenotype assessment;
- d. ability to record clinical data, including recording phenotypic features on permanent media (measurement, photographs, electronic records);
- e. ability to prescribe, assess and interpret the results of diagnostic tests with particular regard to molecular, cytogenetic and enzymatic tests, other additional tests;
- f. ability to prescribe tests and consultations in other medical and non-medical specialties, ability to apply the data thereby obtained for the purposes of establishing diagnoses, prognoses and assessments of the risk of genetic diseases;
- g. ability to talk to families taking due account of psychological and social barriers and other features of such discussions, as well as the principles of non-directive counselling;
- h. ability to diagnose common genetic diseases and syndromes, to differentiate the most common non-genetic syndromes (especially malformation syndromes); knowledge of the principles for diagnosing rare and very rare genetic diseases;
- i. ability to identify and interpret genetic risk on the basis of a diagnosis and the results of lineage analysis;
- j. ability to use computer databases and risk-assessment software;
- k. ability to formulate risks and explain them comprehensibly (orally and in writing) on the basis of the assessment, diagnosis, knowledge of the nature of the disease and other

relevant factors, provide genetic counselling, in particular concerning the disease's characteristics, the possibilities and limitations of treatment and rehabilitation, the reproductive and lifestyle options and preventive-diagnosis procedures for potentially affected family members;

- l. ability to provide and/or supervise prospective care for the affected family in matters pertaining to the genetic nature of the disease;
  - m. ability to keep genetic counselling records in accordance with professional standards and applicable law;
  - n. ability to interpret screening results for the purposes of: genetic counselling, identifying the carriers of specific genetic mutations, assessing genetic predisposition in the context of counselling, prevention, health policy and insurance;
  - o. ability to interpret legal provisions on the diagnosis of genetic diseases and genetic counselling; knowledge of legal provisions and welfare arrangements affecting the quality of life of people with genetic diseases and their families.
4. The duration of the specialty training for graduate doctors (with no specialty) is five years, including teaching, placements and leave.
5. Forms and methods of training:
- a. specialty training courses (compulsory):
    - i. introductory course: "Basics of good medical practice" – duration: 3 days;
    - ii. course: "Basics of clinical genetics" – duration: 7 days;
    - iii. course: "Laboratory diagnostic methods in clinical genetics" – duration: 7 days;
    - iv. course: "Clinical genetics in paediatrics" – duration: 7 days;
    - v. course: "Neurogenetics and clinical genetics of systemic diseases" – duration: 10 days;
    - vi. course: "Oncogenetics - familial cancers" – duration: 7 days;
    - vii. course: "Oncogenetics – leucosis and solid tumours" – duration: 3 days;
    - viii. course: "Clinical genetics of sex and reproduction" – duration: 10 days;
    - ix. course: "Health promotion in clinical genetics" – duration: 2 days;
  - b. recommended specialty training courses (optional);
  - c. internships:
    - i. internship: "Basics of diagnosis and counselling in clinical genetics" - duration: 12 months;  
genetic counselling work (12 months) under the supervision of a specialist combined with training in a cytogenetic laboratory (6 months) and a molecular laboratory (6 months);
    - ii. internship: "Genetics in paediatrics" -duration of internship: 12 months;  
clinical practice in a genetic counselling centre of excellence or reference centre for paediatric genetics under the supervision of a specialist combined with training in one of the following clinical departments or laboratories linked to that centre:
      1. neonatal department – 2 months;
      2. children's department – 3 months;
      3. paediatric surgery department – 1 month;
      4. paediatric orthopaedics department – 1 month;
      5. paediatric haematology department – 1 month;
      6. paediatric neurology department – 1 month;
      7. biochemical laboratory conducting screening and diagnosis of metabolic genetic diseases – 1 month;
      8. cytogenetic laboratory of a reference centre – 1 month;
      9. molecular laboratory of a reference centre – 1 month;
    - iii. internship: "Genetics in gynaecology and obstetrics"  
duration of internship: 12 months;  
clinical practice in a genetic counselling centre of excellence or reference centre

for paediatric genetics under the supervision of a specialist combined with training in one of the following clinical departments or laboratories linked to that centre:

1. an obstetrics department performing non-invasive (including echography) and invasive prenatal diagnosis – 3 months;
2. an obstetrics department performing non-invasive and invasive methods of foetal therapy – 1 month;
3. a gynaecological department treating sex determination and differentiation disorders and paediatric gynaecology and gynaecological endocrinology – 3 months;
4. a gynaecological department diagnosing and treating reproductive failure – 1 month;
5. an affiliated cytogenetic laboratory performing prenatal diagnosis – 1 month;
6. an affiliated biochemical laboratory performing prenatal diagnosis – 1 month;
7. an affiliated molecular laboratory performing prenatal diagnosis – 1 month;

iv. internship: "Oncogenetics"

duration of internship: 6 months;

clinical practice in a genetic counselling centre of excellence or reference centre for paediatric genetics under the supervision of a specialist combined with training in one of the following clinical departments or laboratories linked to that centre:

1. adult oncology department – 1 month;
2. histopathology laboratory – 1 month;
3. molecular laboratory – 2 months;
4. haematological cytogenetic laboratory – 2 months;

v. internship: "Neurogenetics"

duration of internship: 5 months

clinical practice in a genetic counselling centre of excellence or reference centre for paediatric genetics under the supervision of a specialist combined with training in one of the following clinical departments or laboratories linked to that centre:

1. adult neurology department – 1 month;
2. paediatric neurology department – 1 month;
3. neuropathology laboratory – 2 weeks;
4. another neurodiagnostic laboratory – 2 weeks;
5. molecular laboratory – 2 months;

d. training in treatments and medical procedures:

i. independent activities:

1. independently conduct under supervision at least 1000 genetic consultations over the duration of the specialty training, including: 200 concerning paediatric genetics, 100 concerning genetic risk in respect of pregnancies, 200 concerning oncogenetics, 100 concerning neurogenetics, 100 concerning reproductive failure, 100 concerning constitutional karyotype disorders, 100 concerning ophthalmological, dermatological etc. genetic disorders;
2. independently conduct under supervision dysmorphology assessments of 100 children and 50 adults over the duration of the speciality training;
3. independently collect and preserve material for molecular tests from 100 patients, including 20 to isolate DNA from peripheral blood, 10 from other tissues and blocks, 10 to isolate RNA;
4. independently collect material for cytogenetic tests, including 20 for micro and macro methods involving peripheral blood, 10 for dermal fibroblasts, five for other mitotic cells;

ii. activities assisted or supervised by a specialist:

1. hands-on experience of the procedures for extracting chromosomes from cell cultures of different human tissues, the methodology for diagnosing different chromosomal aberrations and recording them in accordance with the international classification;
  2. active participation in the performance of 150 cytogenetic tests, including 20 involving advanced cytogenetic techniques and 10 involving metaphase cells from sources other than peripheral blood and the independent performance of at least 25 routine karyotype tests on cells from different tissues;
  3. hands-on participation in molecular diagnosis using basic DNA analysis techniques in 150 tests, including at least 25 PCR reactions, 10 advanced PCR techniques, 25 PCR-RFLP, 10 polymorphism assessments, 10 applications of other techniques (SSCP, DGGE, etc.), 10 DNA sequencing; independent analysis and presentation of results in at least 20 cases of molecular diagnosis, including at least five different genetic diseases;
  4. hands-on participation in the screening and full laboratory diagnosis of 100 cases of metabolic genetic diseases;
  5. assistance to a medical instructor in the performance of at least 100 USG tests: including 40 screening tests in the first trimester (25 abdominal and intravaginal USGs), 40 in the second and third trimesters and 20 targeted (genetic) USGs in cases where screening has revealed anomalies;
  6. assistance in the performance of at least 20 foetal electrocardiograph tests and at least 10 operations under USG control, at least two of them involving cordocentesis or the placement of intra-uterine shunts; participation and assistance in 20 USG tests on women between the 12th and 24th weeks of pregnancy, including at least six to eight cases of foetal pathology detected by such tests, capacity to use USG to diagnose foetal oedema, measure nuchal translucency, and assistance in 20 invasive procedures;
  7. interpretation of detailed (genetic) results of USG tests on foetuses, foetal echos, cytogenetic and biochemical tests in 100 cases;
  8. performance under supervision, anamnesis, general and neurological testing (including testing of ocular DNA) and presentation of documentation on 30 patients, including 10 children; assistance in psychological testing to reveal symptoms of brain damage resulting from organic changes in the central nervous system of two persons, determination of the IQ of two persons (children); assistance in the performance and interpretation of 10 EEG tests, 10 EMG tests, 10 neuropathology tests and 10 tests involving imaging techniques in cases of suspected genetic diseases; complete interpretation (under the supervision of a neurogeneticist ) of the results of biochemical and molecular tests for genetic neurodegenerative diseases;
  9. assistance in the collection of lineage data, lineage testing, prescription and interpretation of extra clinical, biochemical, histopathological and molecular tests in about 50 cases of different syndromes with a predisposition to family cancers;
  10. participation in the performance of 20 cytogenetic and molecular tests permitting the diagnosis of leucosis, differentiation between cancers and other haematological syndromes; precise diagnosis of a leucosis subtype; assessment of the prognosis, composition of a treatment protocol; monitoring of a course of chemotherapy and a bone-marrow transplant;
- e. forms of autonomous study: study of the literature, participation in the activities of scientific associations, preparation of publications.
6. Methods for assessing knowledge and practical skills:
    - a. seminars;
      - a test or seminar on completion of each specialty course;

- a seminar on completion of each internship on the area of knowledge covered by the internship programme;
  - b. practical skills tests;  
practical tests at the end of each internship;
  - c. assessment of publications;  
assessment and marking of course work or original work by the supervising specialist.
7. State Specialty Examination:  
Speciality training in clinical genetics ends with the sitting of the theoretical and practical parts of State Specialty Examination. The parts of the examination are taken in the following order:
- a. a written examination;
  - b. a practical examination;
    - i. an unassisted case analysis:
      - 1. the physical examination and interview of a patient;
      - 2. planning of the necessary tests;
      - 3. analysis of the available test results;
      - 4. establishment of a diagnosis and differentiation;
      - 5. planning of further action;
    - ii. unassisted performance of cytogenetic and molecular tests and interpretation of the results;
  - c. oral examination.

A doctor who passes the State Specialty Examination qualifies as a specialist in clinical genetics. The diploma is delivered by the Medical Examinations Centre.

The full speciality training programme can be found on the webpage of the Centre for Post-Graduate Medical Training:

[http://www.cmkp.edu.pl/programy\\_pdf/Genetyka%20Kliniczna%20podstawowy%202002.pdf](http://www.cmkp.edu.pl/programy_pdf/Genetyka%20Kliniczna%20podstawowy%202002.pdf)