Study program: Integrated Academic Studies in Medicine

Course title: Clinical Genetics

Teachers: Ivana I. Kavečan, Tatjana B. Redzek Mudrinic, Slobodan D. Spasojević

Course status: elective

ECTS Credits: 3

Condition: -

Course aim

The primary aim of the elective course of Clinical Genetics is to inform medical students about the possibilities of applying clinical genetics in primary, secondary and tertiary health care; acquiring knowledge about the possibilities of applying new molecular genetics technologies in clinical genetics and clinical practice; introduction to the importance of clinical recognition, diagnosis and treatment of hereditary diseases, including rare hereditary diseases. Aquiring knowledge on the importance of prenatal diagnosis including preimplantation genetic testing. Aquiring knowledge on the importance of newborn screening.

Expected outcome of the course

Taking the elective Clinical Genetics course, students will get knowledge on approuch to patients who suffer from inherited diseases. Students will be informed on the importance of etiology, pathogenetic mechanisms, clinical presentation and treatment options for hereditary diseases, including diseases with low incidence (rare diseases). Students will become informed about the importance of dysmorphological examination. Particular attention is paid to modern approaches to the detection and treatment of hereditary diseases, as well as the proper differential diagnosis and additional diagnosis of hereditary diseases, including rare congenital diseases of metabolism. Students will be informed with the importance of prevention of hereditary diseases, including the possibilities of preimplantation genetic testing. Students will be introduced to newborn screening.

Course description

Theoretical education

- Inherited diseases
- Basic principles of hereditary diseases
- Inherited diseases with non-traditional pattern of inheritance
- Chromosomal abberations
- Monogenic diseases
- Multifactorial diseases
- Mitochondrial pattern of inheritance
- Expansion of repetitive trinucleotides
- Genetic imprinting and uniparental dysomia
- Epigenetics
- Dysmorphology
- Congenital anomalies
- Mutations
- Genetic counseling
- Prenatal diagnosis
- Non-invasive prenatal diagnosis
- Invasive prenatal diagnosis
- Preimplantation genetic testing
- Genetic aspect of infertility
- Ethical problems in clinical genetics
- New technologies in molecular genetics including next-generation sequencing
- Modern therapeutic approach for persons affected with hereditary diseases
- Neonatology and genetics
- Inherited metabolic diseases that are available for treatment
- Inherited metabolic diseases not available for treatment
- Personalized medicine
- A multidisciplinary approach in clinical genetics

Practical education

- A family medical history in clinical genetics
- Standard pedigree symbols, pedigree analysis
- Physical examination including dysmorphological examination of the patient, introduction to the terminology of clinical dysmorphology
- Case reports of numerical chromosomal anomalies
- Case reports of structural chromosomal anomalies
- Case reports of monogenic diseases
- Case reports of mitochondrial diseases
- Case reports of uniparental dysomia
- Case reports of neurological hereditary diseases
- Case reports of congenital anomalies
- Case reports of congenital metabolic disorders
- Introduction to non-invasive prenatal diagnostic methods
- Introduction to invasive methods of prenatal diagnosis
- Introduction to preimplantation genetic testing
- Isolation of DNA from a blood sample, amnion, fetal blood and skin
- Introduction to MLPA technology
- Karyotype analysis by standard techniques and other techniques
- Introduction to the detection of chromosomal microdeletions
- Introduction to the detection of mutations of genes responsible for cystic fibrosis
- Introduction to microarray technology
- Introduction of enzyme replacement therapy
- Introduction to the use of substrat inhibitory treatment and enzyme cofactor treatment
- Introduction to prenatal diagnosis
- Introduction to newborn screening
- Introduction of personalized medicine's possibilities

Literature

Required

1. Firth H, Hurst J. Oxford Desk Reference Clinical Genetic and Genomics, 2nd Ed. Oxford University Press 2017.
2. Nussbaum RL, McInnes RR, Willard HF. Thompson and Thompson Genetics in Medicine, 8th Ed. Elsevier Science Health Science 2015.

Number of active classes	**Theoretical classes:** 30	**Practical classes:** 15

Teaching methods

Lectures, practice Medical history, physical, dysmorphological examination of persons affected by hereditary diseases, differential diagnostic and therapeutic aspects in clinical genetics with case reports; Presentation of the Cytogenetic Laboratory; Molecular Genetic Laboratory; Medicogenetic Service for Family Planning and for Medical Genetics – Service for Medical Genetics.

Student activity assessment (maximally 100 points)

Pre-exam activities	**points**	**Final exam**	**points**
Lectures	20	written exam	20
Practice	30	oral exam	30
Colloquiums		.....	
Seminars			