Study program: Integrated academic studies in medicine

Type and level of the study program: integrated academic studies

Course title: Clinical genetics (M3-CGEN)

Teacher: Jadranka D. Jovanović-Privrodski, Aleksandra R. Doronjski, Nada V. Konstantinidis, Ljubica L. Georgijević, Dragan J. Katanić, Jovanka L. Kolarović, Ivana I. Kavečan, Nenad A.Barišić

Course status: ELECTIVE

ECTS Credits: 3

Condition: -

Course aim

Students get acquainted with specific features of clinical genetics, clinical manifestations, differential-diagnostic procedures and treatment of hereditary diseases. Students will be able to work with specific populations as general practitioners.

Expected outcome of the course:

Students will acquire knowledge on etiology, pathogenesis, clinical picture and treatment of hereditary diseases, with emphasis on the specifics population groups with hereditary diseases. Particular attention is paid to the importance of preventive measures and procedures, as well as to differential diagnosis. Students will get acquainted with prenatal diganostics and the possibilities of prevention.

Appropriate approach and communication with patients with hereditary diseases and their families. Specific methods of obtaining medical history, physical examination and specific therapy procedures in this patient population. Specific approach in primary care institutions when working with patients with hereditary diseases.

Course description

Theoretical education

- Clinical Genetics (Introduction)
- Ethics in clinical genetics
- DNA as the hereditary materia
- Gene (gene structure)
- Human chromosomal material, karyotype, karyogram
- Hereditary disease: definition, classification, prevalence
- Congenital anomalies
- Monogenic diseases; autosomal dominant inheritance-AD; autosomal recessive inheritance AR
- Monogenic diseases XR inheritance, XD inheritance; characteristics of most prevalent diseases
- Mitochondrial diseases, inheritance, most prevalent diseases
- Autosomal chromosomes and related diseases
- Sex chromosomes and diseases
- Multifactorial diseases (inheritance, most prevalent diseases)
- Genetic counselling, genetic information
- Prenatal diagnostics - classification and importance
- Noninvasive prenatal diagnosis
- Invasive prenatal diagnosis
- Treatment of hereditary diseases

Practical education: exercises, other forms of education, research related activities

- Medical history (obtaining and importance)
- Genealogy (each student makes his DNA family tree)
- Physical examination
- Minor malformation score (MMS) (each student calculates his own MMS)
- Case reports: hereditary diseases; autosomal chromosome diseases; Down syndrome; Edwards syndrome; Pat syndrome; Wolf syndrome
- Case reports: hereditary diseases; sex chromosome diseases; Klinefelter syndrome, Turner syndrome, fragile X syndrome
- Case reports: monogenic diseases; neurofibromatosis; Marfan syndrome; spinal muscular atrophy; haemophilia; Achondroplasia; etc.)
- Case reports: genetic counselling
- Invasive prenatal diagnostics
- POOSUM (hereditary disease diagnostic software)
- karyotype G analysis technique (microcsopy)
- Syndromatology (film)

Literature

Compulsory

1. Turnpenny P, Ellard S. Emery's Elements of Medical Genetics, 15th Edition. Elsevier, 2017.
2. Nussbaum R, McInnes R, Willard HF. Genetics in Medicine, 8th Edition. Thompson & Thompson, 2015.

Number of active classes				Other:
Lectures: 30	Practice: 15	Other types of teaching:	Research related activities:	

Teaching methods

Lectures and practical work.

Student activity assessment (maximally 100 points)

Pre-exam activities	**points**	**Final exam**	**points**
Lectures	20	Written	20
Practices	30	Oral	30
Colloquium		.....	
Essay			