Study program: Integrated academic studies in Medicine

Type and level of the study program: integrated academic studies

Course title: Human genetics (M1-HUGE)

Teacher: Mihajla R. Đan, Nataša S. Vučinić

Course status: compulsory

ECTS Credits: 6

Condition: -

Course aim

This course has been designed to introduce fundamental concepts and technological advances in the study of human genome as well as genetic mechanisms of hereditary diseases. Each of the major subspecialties will be addressed: cytogenetics, molecular genetics, biochemical genetics, clinical genetics, reproductive and prenatal genetics and genetic counselling. Genetic variability provides the basis for the development of both, rare diseases and common disorders.

Expected outcome of the course:

Students will be able to use basic genetic entities and recognize the importance of genetics in modern science. They will get knowledge about: morphological and functional organization of chromosomes and mutation mechanisms. Using examples they will apply Mendel laws, understand intra and interloci gene interactions. They will be able to predict possible mechanisms of inheritance and to construct genealogy. They will understand and differ basic techniques of molecular genetics in prenatal diagnostics, forensics and gene therapy. After fulfilling pre-exam and exam obligations students will be able to: differ levels of structural and functional organization of human genome; identify mechanisms of gene expression; comprehend processes of genetic experimentation leading to determination of the etiology of hereditary diseases of various complexity; explain the methodology of basic techniques in molecular genetics in prenatal diagnosis and forensics; use available internet sources and professional literature.

Course description

Theoretical education

Introduction to human genetics. Structure of nucleic acids; Gene expression and gene regulation. Human genome structure; Morphologic and mollecular organization of chromosomes; Cell division. Gametogenesis; Basic laws of heredity. Autosomal dominant and autosomal recessive inheritance. Genealogy analysis; Intralocus and interlocus gene interactions; Sex determination. Sex and holandric inheritance; Polygenic inheritance. Multifactorial and complex hereditary diseases; Mutations, reparation and DNA recombinations; Variations in number and structure of chromosomes. Hereditary diseases caused by chromosomal aberrations; Mollecular markers in human genetics. Prenatal and preimplantation diagnosis of genetic diseases.

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

Nucleic acids and gene expression; Structural and mollecular organization of chromosomes; Cell division; Basic laws of heredity; Genealogy analysis; Gene interactions. Multiple alleles and blood groups; Gene interactions. Partial dominance. Codominance. Epistasis; Sex determination. Sex-related characteristics; Chromosomal aberrations; Chromosomal structure mutations; Multifactorial inheritance; Molecular markers in human genetics.

Literature

Compulsory

1. Turnpenny P, Ellard S. Emery's elements of medical genetics. Elsevier; 2009.

Additional

1. Lewis R. Human genetics. UK: Cambridge University Press; 2007.
2. Strachan T, Read A. Human molecular genetics. 4th ed. USA: Garlan Science; 2011.

Number of active classes				Other:
Lectures: 45	Practice: 30	Other types of teaching:	Research related activities:	

Teaching methods: lectures, practice

Student activity assessment (maximally 100 points)

Pre-exam activities	**points**	**Final exam**	**points**
Lectures		Written	65
Practices	5	Oral	
Colloquium	30	.....	
Essay			