UNIVERSITY OF NOVI SAD
FACULTY OF MEDICINE

Study program: Integrated Academic Studies in Medicine

Course title: Biology with Human Genetics

Teacher: Đan R. Mihajla, Vučinić S.Nataša

Course status: compulsory

ECTS Credits: 8

Condition: -

The aim of the course is to acquaint students with the organization and structure of the cell and cell organelles, the organization of the human genome and the expression of the human genome, as well as with the genetic mechanisms of hereditary diseases. Throughout the course, the student will use numerous sources of information through various forms of teaching activities and gain new knowledge of the fundamental concept and technological advances in human genome research, and will also gain insight into the human microbiome. The aim of the course is to understand and adopt the processes and mechanisms of transferring of the structure and expression of genetic information at the levels of molecules, chromosomes, organisms and populations.

Expected outcome of the course:

After completing the course, the student will understand theories about the evolution of life, learn the structure of the cell and cell organelles, and distinguish between prokaryotic and eukaryotic cells as well as plant and animal cells. They will be able to recognize the importance of certain organelles in the transmission and regulation of gene expression. The student will understand basic genetic concepts and recognize the importance of genetics in modern science. They will learn about chromatin structure, morphological and functional organization of chromosomes. They will clearly distinguish between the stages of meiosis and understand the importance of cell division in transmission genetics. Through examples, they will apply Mendel's laws, understand the intra and inter locus interactions of genes. Students will anticipate possible mechanisms of inheritance and accurately construct pedigree based on given data. They will understand the mechanisms of mutations, and the mechanisms of DNA repair. They will understand and differentiate basic molecular genetics techniques in prenatal diagnosis, population genetics and forensics and understand the principles of gene therapy. After successfully completing the pre-exam and exam obligations, the student will be able to distinguish between levels of structural and functional organization of the human genome; identify mechanisms of regulation of gene expression; understand the processes of genetic experiments that explain causes of hereditary diseases of varying complexity and etiology; explain the methodology of basic molecular genetics techniques in prenatal diagnosis, population genetics and forensics; use internet sources and professional literature with understanding.

Course description

Theoretical education

Evolution of life and the formation of cells and nucleic acids. Cell structure, cell organelles, transport trough the cell membrane. Human microbiome. Nucleic acid structure, DNA replication and types of RNA molecules. Gene expression and control of gene expression. Molecular organization of chromosomes, organization of the human genome. Cell cycle and cell division, gametogenesis, causes of chromosome non-disjunction. Basic principles of inheritance, Mendel's laws, classification of genetic diseases, formation of pedigree. Extension of Mendel's laws: incomplete dominance, codominance, multiple alleles, mitochondrial inheritance. Extension of Mendel's laws: expressivity and penetrance, pleiotropy, phenocopies, genocopies, lethal alleles, linked genes, gene interactions. Autosomal dominant diseases, autosomal recessive diseases. Polygenic, multifactorial and complex diseases. Sex chromosomes and sex-related traits, traits influenced by sex. Sex determination in humans and disorders of gender differentiation. Chromosome aberrations: structural and numerical and analysis of selected syndromes caused by aberrations. Uniparental disomy and gene imprinting. Gene mutations, recombinations, DNA repair mechanisms and diseases associated with malfunctioning of DNA repair mechanisms. Molecular methods in human genetics. Prenatal and preimplantation diagnosis of genetic diseases; Genetic counseling. Possibilities of gene therapy.

Practical education

Nucleic acids and gene expression; Structural and mollecular organization of chromosomes; Cell division; Basic laws of inheritance; Pedigree analysis; Gene interactions. Multiple alleles and blood groups; Gene interactions. Partial dominance. Codominance. Epistasis; Sex determination. Sex-related traits; Chromosomal numeric aberrations; Chromosomal structural aberrations; 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, 12th edition Mc Graw-Hill Education, New York, 2018.
2. Alberts B, Johnson A, Lewis J, Morgan D, Raff M, Roberts K, Walter P. Molecular Biology of the Cell, Sixth Edition. Garland Science, Taylor & Francis Group, New York, US, 2015.

Number of active classes	**Theoretical classes:** 45	**Practical classes:** 30

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			