Study program: REPRODUCTIVE BIOLOGY

Course title: Human genome

Teacher: Nevena Veličković

Course status: obligatory

ECTS: 5

Requirements:

Course objectives

The aim of this course is to introduce students to the human genome organization, distribution and function of genes and genetic mechanisms of monogenic and complex genetic diseases. The students will achieve basic laboratory techniques and get introduced to all available human genome databases, in order to acquire new knowledge on technological advances and discoveries, and mechanisms of epigenetic control.

Learning outcomes

After successful fulfilling of pre-exam and exam obligations student can:
- differ levels of structural and functional human genome organization
- with the understanding to describe the idea, history and technology of the Human Genome Project
- understand the mechanisms of pathogenic mutations
- describe and implement the methodology for gene identification in monogenic and complex genetic diseases
- understand epigenetic phenomena
- use internet resources and literature in the field of human genomics

Syllabus

Theoretical instruction

The concept of gene, genome, genomics and new sequencing technologies (NGS). Human Genome Project. The human genome organization. The organization, distribution and function of the protein coding genes of the human genome. The organization, distribution and function of RNA genes of the human genome. Noncoding regions of the human genome, repetitive DNA, transposable elements. Human mitochondrial genome. Human gene expression. Human genetic variability. Pathogenic mutations. DNA polymorphism. Dynamic mutations. Epigenetic mechanisms. Epigenetic reprogramming in the early embryonic stages. Application of information of human genome, preimplantation and prenatal genetic diagnostics, forensics. Principles and examples of gene therapy in the human population. Cancer genetics and genomics.

Teaching laboratory, Practical laboratory

Practical work. Karyotyping. Pedigree analysis. Molecular markers: the selection of marker systems in diagnostics, population genetics and forensics. Use of internet sources with information on the human genome organization and function: NCBI, OMIM, GENOME.

Other forms of teaching (laboratory work). Extraction of DNA from human material. PCR amplification of the genes and gene fragments, analysis of polymorphism by RFLP and/or differences in the length of the PCR product. Determination of genotype.

Literature

1. Stachan T, Read AP. Human Molecular Genetics 4 Garland Publishing, UK, 2011.
2. Turnpenny P. Ellard S. Emery's elements of medical genetics, 12th Ed., Elsevier, Churchill Livingstone, 2005.
3. Relevant scientific papers in the field.

Weekly teaching load

Lectures: 2	Practical lectures:1+1+0

Teaching methods

lectures, practical lectures, laboratory work, seminar, tuition

Evaluation of knowledge (maximum score 100)

Pre-exam obligation	Points	**Final exam**	Points
Student engagement in lectures		Test/Written exam	
Practical laboratory – computer work	10	Oral exam	60
Practical laboratory – lab report	30		