Course Unit Descriptor

Study Programme: Reproductive biology

Course Unit Title: Human genome

Course Unit Code: RB05

Name of Lecturer(s): Prof. Dr. Mihajla Djan

Type and Level of Studies: Master's studies

Course Status (compulsory/elective): elective

Semester (winter/summer): winter

Language of instruction: English

Mode of course unit delivery (face-to-face/distance learning): face-to-face

Number of ECTS Allocated: 5

Prerequisites:

Course aims:

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.

Learning outcomes

After successful fulfilling of pre-exam and exam obligations student can: differ levels of structural and functional human genome organization and understand the mechanisms of pathogenic mutations, and explain the methodology for gene identification in monogenic and complex genetic diseases.

Syllabus

Theoretical instruction

The human genome organization. The organization, distribution and function of RNA genes of the human genome. The organization, distribution and function of the protein coding genes of the human genome. Noncoding regions of the human genome. Human mitochondrial genome. Pathogenic mutations. Identification of the genes in monogenic diseases. Identification of genes determining complex genetic diseases. Molecular pathology. Cancer genetics. Epigenetic mechanisms. Epigenetic reprogramming in the early embryonic stages.

Teaching 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. Strachan T., Read AP. Human Molecular Genetics 4th edition. Garland Publishing, New York, USA. 2011. 2. Lewis R. Human Genetics: Concepts and Applications, 9th edition, McGraw–Hill Primis, USA, 2009.

3. Allis CD, Jenuwein T, Reinberg D, Caparros ML. Epigenetics. Cold Spring Harbour Laboratory Press, New York, USA, 2007.

Tollefsbol T. Handbook of Epigenetics The New Molecular and Medical Genetics, Elsevier, USA, 2011.
Turnpenny P, Ellard S. Emerijevi osnovi medicinske genetike. XIII izdanje. Datastatus, Beograd, Srbija. 2009..

Weekly teaching load	Lectures: 2	Teaching laboratory: 1	Other forms of teaching: 2
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		Written exam			
Seminar	Up to 10	Oral exam	Up to 60		
Tests					
Practical laboratory	Up to 30				