Study program: Integrated academic studies of Pharmacy

Type and level of the study program: integrated academic studies

Course title: BIOLOGY WITH HUMAN GENETICS (PhI-BHG)

Teacher: Nataša S. Vučinić

Course status: compulsory

ECTS Credits: 6

Condition: -

Course aim

The aim of this course is to introduce students to the organization and expression of the human genome and the genetic mechanisms of hereditary diseases. The student will, during the course through various learning activities, use many sources of information and gain knowledge on fundamental concepts and technological advances in the research of human genomes. Furthermore, the aim is to understand and adopt processes and mechanisms of transmission of the structure and expression of genetic information at the molecular level, chromosome, organism and population.

Expected outcome of the course:

Students will use basic understanding of genetic concepts and recognize the importance of genetics in modern science. Become informed about the chromatin structure, morphological and functional organization of chromosomes. They will clearly distinguish the stages of meiosis and understand the importance of cell division in a transmission detection genetics. Through examples they will apply Mendel's laws, understand intra and interlocus interactions between genes. Also, they will predict potential mechanisms of inheritance and accurately construct family trees from the data, understand the mechanisms of mutations, mutagenic effects of the principles and mechanisms of DNA repair. They will understand and distinguish the basic techniques of molecular genetics in prenatal diagnosis, population genetics and forensics. They will understand the principles of gene therapy.

Course description

Theoretical education

1. Introduction to Human Genetics. The development and significance of human genetics. 2. Nucleic acid. The structure of DNA and RNA. DNA replication. Gene expression. 3. Molecular organization of the cell. Prokaryotic and Eukaryotic cells. Structure and function of cell organelles. 4. Molecular organization of chromosomes. 5. Cell cycle. Mitosis, meiosis and gametogenesis. Chromosome analysis. 6. Laws of inheritance. Mendel's rules of inheritance. Pedigree. 7. Non-Mendelian inheritance: Incomplete dominance, codominance, multiple alleles, polygenic traits, lethal alleles. 8. Penetrance and expressivity, pleiotropy, genetic anticipation, genome imprinting. 9. Functional organization of the human genome. The composition of the human genome, the organization and distribution of genes, family genes, repeated genes, mobile genetic elements, expansion of trinucleotide repeats. The autonomy of the mitochondrial genome. 10. Mutations, repair and recombination. Genotoxic effects of chemical, physical, biological, mutations on molecular level, chromosomal aberrations, repair, recombination. 11. Molecular markers. DNA polymorphism and genetic markers in the human population. Mapping and sequencing the human genome. Cloning of the gene. Gene Therapy. 12. Prenatal diagnosis of genetic diseases. Techniques which are used in prenatal and preimplantation diagnostics. 13. The inheritance of diseases. Diseases that are inherited autosomal dominant and autosomal recessive diseases that are inherited multifactorial, sex linked inheritance and sex influenced disorders

Practical education

1. Microscopes and microscopy of size and shape of cells and nuclei. 2. Microscopy of cell membrane and cytoplasmic organelles. 3. Microscopy morphology and classification of chromosomes, interphase nucleus, chromatin, karyotype. 4. Nucleic acids. Purine and pyrimidine bases, nucleosides and building blocks of nucleotides, the structure and function of DNA and RNA (tasks). 5. Cell division and gametogenesis. Cell cycle, interphase, mitosis, meiosis, spermatogenesis and oogenze flow. 6. Microscopy of mitosis, the recognition phase. 7. Basic laws of inheritance. Definition of basic genetic concepts. Mendelian rules of inheritance. Crossing and mode of inheritance of certain traits through given problems. 8. Analysis genealogy. Symbols of a family tree, genealogy interpretation, forming genealogy, determining the mode of inheritance of properties on the basis of the genealogy. 9. Deviation from Mendel's rules. The ratio of full domination of codominance and partial dominance, epistasis. 10. Properties associated with sex chromosomes X and Y. Sex linked inheritance and sex influenced disorders. 11. Multiple alleles. Multiple alleles - ABO and Rh blood group system. 12. Changes in the number and structure of chromosomes. Numerical aberrations: polyploidy and aneuploidy. Structural chromosomal aberrations: duplications, deletions, inversions and translocations. 13. Population genetics. Population structure, calculating the frequency of alleles and genotypes. 14. Isolation DNA electrophoresis. Methods for DNA isolation and electrophoresis types. 15. Molecular markers. protein markers, molecular markers (RFLP, PCR), sequencing. 16. 16 Forensics. Examples, use and importance.

Literature

Compulsory

1. Strachan T, Read AP. Human Molecular Genetics, 4th edition. Garland sciences, UK, 2011.
2. Turnpenny P, Ellard S. Emery's Elements of Medical Genetics, Churchill Livingstone, 2007.

Optional

1. Thompson&Thompson. Genetics in Medicine. Nussbaum Saunders Elsevier, 2007.
2. Lewis R. Human Genetics. McGraw-Hill Science, 2007.

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

Teaching methods: Theoretical and practical education

Student activity assessment (maximally 100 points)

Pre-exam activities	**points**	**Final exam**	**points**
Lectures		Written	67
Practices	3	Oral	
Colloquium	2x15		
Essay			