

# Study program: Integrated Academic Studies in Pharmacy

# **Course title: Biology with Human Genetics**

Teacher: Nataša S. Vučinić

Course status: compulsory

#### ECTS Credits: 6

Condition: -

# Course aim

The objective of the course is to introduce students to the evolution of genetic material and cells to explain the organization and structure of the cell and cell organelles, as well as the organization of the human genome and the expression of the human genome. Aim ot this course is to explain 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. Also the object of the course is to understand processes and mechanisms of transfer, structure and expression of genetic information, as well as different levels of controlling gene expression. Understanding the importance of genetic mechanisms of drug resistance as well as insight into the human microbiome.

### Expected outcome of the course:

After completing the course, the student will understand theories of life, learn the structure of the cell and cell organelles, and will be able to make distinction between prokaryotic and eukaryotic cells, as well as plant and animal cells. Students will recognize the importance of certain organelles in the transmission and regulation of gene expression. They will be able to use basic genetic concepts and recognize the importance of genetics in modern science. They will learn about the structure of chromatin, the morphological and functional organization of chromosomes. They will clearly differentiate between stages of meiosis and understand the importance of cell division in transmission genetics. Through examples, they will apply Mendelian laws, understand the intra and inter locus gene interactions, as well as deviations from Mendelian inheritance. They will be able to determine possible mechanisms of inheritance and interpret deviations from Mendelian laws of inheritance, and accurately construct pedigree based on given data. They will be introduced to sex limited traits and disorders that can occur during sex limited inheritance, as well as sex influenced traits. Students will understand causes and types of mutations, and the possible mechanisms of DNA repair, as well as the various mechanisms of bacterial cell resistance to antibiotics. They will see the importance of selective pressure and the potential threat of a returning to the pre-antibiotic era. They will understand basic molecular genetics techniques.

### **Course description**

### Theoretical education

The evolution of life and the formation of cells and nucleic acids. 2. Cell structure, cell organelles, transport. 3. Nucleic acid structure, DNA replication, and types of RNA molecules. 4. Gene expression and control of gene expression 5. Molecular organization of chromosomes, organization of the human genome. 6. Cell cycle and cell division, gametogenesis, causes of chromosome disjunction. 7. Basic principles of inheritance, Mendel's laws, classification of genetic diseases, formation of pedigree.
Beyond Mendel's laws: incomplete dominance, codomination, multiple alleles, mitochondrial inheritance. 9. Beyond Mendel's laws: penetrance and expressivity, pleiotropy, phenocopies, genocopies, lethal allele combinations, linked genes, gene interactions.
Autosomal dominant diseases, autosomal recessive diseases. 11. Sex chromosome aberrations: structural and numerical and analysis of selected syndromes caused by aberrations 14. Mosaicism, Chimerism, Gene Mutations, DNA reparation mechanisms, and diseases associated with impared DNA reparative mechanisms. 15. Human microbiome, mechanisms of genetic resistance of bacteria to antibiotics, importance of selective pressure and the potential threat of a returning to the pre-antibiotic era.

### Practical education

1. Nucleic acids, structure and classification 2. Gene expression 3. Chromosome morphology and classification, karyotype, karyogram. 4. Cell cycle, cell division and gametogenesis. 5. Basic laws of inheritance. Defining basic genetic concepts. Mendel's rules of inheritance. Crosses and inheritance of certain traits through problems. 6. Pedigree construction and analysis; determination of inheritance pattern based on Pedigree. 7. Beyond Mendel's Laws 1 8. Beyond Mendel's Laws 2. 9. Gendered and gender-related traits. 10. Pedigree analysis of sex-linked traits 11. Atypical Chromosome Number 12. Atypical Chromosome Structure. 13. Population genetic structure and quantitative traits 14. Molecular markers (RFLP, PCR), sequencing. 15. Forensics. Examples, uses and significance.

Literature

# Compulsory

- 1. 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.
- 2. Lewis R. Human Genetics, 12<sup>th</sup> edition Mc Graw-Hill Education, New York, 2018.

Optional

1. Thompson&Thompson. Genetics in Medicine. Nussbaum Saunders Elsevier, 2007.

Number of active classes	<b>Theory:</b> 45	Theory: 45Practice: 45	
Teaching methods: Theoretical and pra	ctical education		
Student activity assessment (maximally	100 points)		
Pre-exam activities	points	Final exam	points
Lectures		Written	65
Practices	5	Oral	
Colloquium	2x15		
Essay			