Course Information Sheet (in the structure according to Decree No. 614/2002 Coll.)

University: University of Prešov in Prešov	
Faculty: Faculty of Humanities and Natural Sciences	
Code: 2BIO/EROSJGENE/22	**Title of Course:** Genetics
Type, scope and method of educational activities: Compulsory course **Form of Study:** Lecture, practical seminar **Number of contact hours:** 2/0/1 **per week:** 2/0/1 **per level/semester:** 26/0/13 **Study method:** combined **Student workload: 150 hours** Direct education: 30 hours Self-study: 50 hours Guided work: 70 hours	
Number of credits: 5	
Semester: 3rd semester	
Degree/Level: 1st degree	
Prerequisities: -	
Grading Policy (Assessment/Evaluation): Exam Completion of the course is assessed by a final examination. During the semester, on the basis of the teacher's instructions, the student prepares, presents and submits a seminar paper according to the assigned topic in the scope of 20 pages (introduction, core, conclusion, list of bibliographic references). During the semester, the student will take written examinations in genetic terminology and genetic examples, which account for 30% of the grade. The final written (oral) examination accounts for 70% of the final grade. A grade of A: 100 – 90 % of the marks, B: 89 – 80 % marks, C: 79 – 70 % marks, D: 69 – 60 % marks, E: 59 – 50 % marks. A student scoring less than 50% will be graded FX. The final grade will be calculated as the average of the grades on the term paper, the written test on genetic terminology, the examples, and the final exam. Attendance at lectures and seminars (direct instruction) is compulsory. A student may have a maximum of 2 absences excused on the basis of a medical certificate. For absences, the student will be given make-up assignments or undergo counselling. In case of unexcused absences or a greater number of absences, no credits will be awarded to the student.	
Aims and Objectives: **Knowledge gained:** The student will be able to: - define and explain in own words concepts from genetic terminology, - describe the structure and types of DNA and RNA, - characterize and explain DNA replication, transcription, translation, properties of the genetic code and solve given problems, - describe the cell cycle, explain the different phases of the cell cycle and regulatory mechanisms, - describe amitosis, mitosis, meiosis, apoptosis and necrosis, - define and explain in their own words the concept of chromosome, the number and structure of chromosomes, the differences between prokaryotic and eukaryotic chromosomes, - characterise and explain modes of inheritance - monogenic, multifactorial, polygenic, extra-nuclear inheritance,	

- define the terms mutation and mutagen, correctly classify mutations, give examples of mutations,
- explain mutagenesis, characterize the symptomatology of selected mutations and assess the difference between numerical and structural chromosomal aberrations,
- describe Mendel's rules of inheritance - laws, give examples, solve given problems,
- define Morgan's rules - gene linkage, forms, phases, examples, solve given problems,
- explain the genetic basis of biochemical diseases, hereditary enzyme deficiencies in humans and animals,
- clarify the meaning of population genetics, classify the genetic structure of a population and describe the law of genetic equilibrium in their own words in the context of autogamous and panmictic population models,
- describe the Hardy-Weinberg law of genetic equilibrium (give formulas for calculating allele and genotype frequencies, solve given problems),
- characterize the investigative methods used in genetics - hybridological, genealogical, gemelilogical, cytogenetic, molecular-genetic; be able to construct a pedigree.

Skills acquired:

The student will be able to:
- independently prepare a term paper on a selected topic in the field of genetics,
- prepare a PowerPoint presentation,
- present the seminar paper in class and discuss the topic.

Competences acquired:

The student will be able to:
- use and apply the acquired knowledge in the context of the content of other subjects,
- understand interrelationships and contexts,
- hierarchically organise the acquired knowledge and facts in genetics,
- look for causal relationships in acquired knowledge and processes, thus developing logical thinking,
- recognise clinical manifestations, causes of genetic pathologies, consequences of pathologies for human life and health,
- to search for information in the field of genetics in professional literature and information media, to work with relavant information,
- develop reading literacy in the study of professional texts in the field of genetics,
- creatively solve given tasks, point out the causes of problems and propose solutions,
- use the acquired knowledge, skills and abilities in further studies and apply them in practice.

Syllabus/Indicative Content:

Introduction to genetics. Basics of genetic terminology.

Molecular basis of genetic information - structure and types of DNA and RNA.

Replication, transcription, translation of DNA, genetic code (examples, solving of tasks).

Laws of cell division - cell cycle, cell cycle regulation, cell differentiation. Programmed cell death - apoptosis, unprogrammed cell death - necrosis, cell senescence.

Laws of reproduction, types of reproduction - asexual, sexual reproduction. Gametogenesis, spermatogenesis, apomixis, in vitro fertilization.

Chromosomal basis of inheritance - structure and number of chromosomes, prokaryotic and eukaryotic chromosomes.

Heredity of organisms - monogenic inheritance, multifactorial inheritance, polygenic inheritance, extra-nuclear inheritance.

Mendelian rules of inheritance - laws, examples, solving problems. Morgan's rules - gene linkage, forms, phases, examples, solving problems.

Mutations - classification of mutations (spontaneous, induced, mitochondrial, somatic, genetic), mutagens, mutations and environment.

Chromosomal aberrations - numerical, structural, causes of chromosomal aberrations.
Genetics of biochemical processes - biochemical diseases and their nature, hereditary enzyme deficiencies in humans and animals, disorders of carbohydrate, protein, lipid, haemoglobin metabolism.
Gene regulation of ontogenesis - regulation during zygote budding, cell differentiation and embryonic induction, sex ontogeny in mammals, humans, etc.
Morphological developmental defects - natural, pharmaceutical, industrial, agricultural, metabolic teratogens. Drugs. Mutants with malformation and lethal effect.
Genetically determined pathologies - numerical aberrations of autosomes, gonosomes, structural aberrations of chromosomes.
Population genetics - genetic structure of population, model of autogamous and panmictic population, Hardy-Weinberg law of genetic equilibrium (examples, solving of given problems), gene pool of population, migration, adaptive value and genetic load of population.
Investigative methods used in genetics - hybridological, genealogical, gemelilogical, cytogenetic, molecular-genetic and others.
Prenatal diagnostics - invasive and non-invasive methods of prenatal diagnostics, FDD-MB method.
Epigenetics - DNA methylation, histone modification, epigenetic inheritance.
Metabolomics - investigation of metabolic pathways, targeted metabolite analysis.
Genomics - structural and functional genomics. Personal and population genomics.

Suggested readings:

PORÁČOVÁ, J., ŠUTIAKOVÁ, I., PETRÁŠOVÁ, D., a kol. 2006. Základy genetiky pre študentov vysokých škôl prírodovedného zamerania. Prešov: FHPV PU v Prešove. 267 s. ISBN 80-8068-455-3.

ROSYPAL, S. 2006. Úvod do molekulární biologie. Díl první. Brno: Tiskárna Těchov, Blansko. 289 s. ISBN 80-902562-5-2.

SRŠEŇ, Š., SRŠŇOVÁ, K. 2005. Základy klinickej genetiky a jej molekulárna podstata. 4. prepracované a rozšírené vydanie. Martin: Osveta. 445 s. ISBN 80-8063-185-9.

KOČÁREK, E. 2008. Genetika. Mníšek pod Brdy: Scientia. 212 s. ISBN 9788086960364.

NUSSBAUM, R. L., MCINNES, R. R., WILLARD, H. F. 2004. Klinická genetika. Praha: Triton. 426 s. ISBN 88-7254-475-6.

OTOVÁ, B., MIHALOVÁ, R. 2013. Základy biológie a genetiky človeka. Praha: Karolinum, 228 s. ISBN 978-80-2462-109-8.

ČELLÁROVÁ, E. a kol. 2001. Príklady zo všeobecnej genetiky. Košice: PF UPJŠ. 126 s. ISBN 80-7097-460-5.

PRITCHARD, D. J. 2021. Základy lékařské genetiky. 2. vyd. Praha: Galén. 242 s. ISBN 978-80-7262-449-2.

TANERI, B. et al. 2020. Human Genetics and Genomics. Germany: Wiley – VCH Verlag GmbH & Co. 163 p. ISBN 978-3-527-68263-8.

Language of Instruction: Slovak language

Other course information: -

Grading history

Total number of assessed students:

A	B	C	D	E	FX

Lecturer/Instructor:

prof. MVDr. Janka Poráčová, PhD., MBA, lecturer, examiner, practitioner
doc. RNDr. Marta Mydlárová Blaščáková, PhD., lecturer, examiner, practitioner

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