Name of university, Name of faculty: Trnava University

Faculty of Health Care and Social Work

INFORMATION SHEET OF THE SUBJECT

Code: LMbD22	**Name:** Human genetics	
Cover: Department of Theoretical Disciplines and Laboratory Investigation Methods		
Type of educational activity: Lecture, Practise **Scope of educational activity (in hours):** **Weekly:** 1/2 **For term of study:** ZS 23,46 **Method of educational activity:**	**Number of credits:** 4	**Recommended semester:** WT **Study grade:** 1. PO

Recommended semester	Study programme
2.year WT	Laboratory Medicine (D1-LVMvZ-22)

Underlie subjects: LMbD04 - Biology I., LMbD11 - Biology II.

Conditions for passing the course:
Method of evaluation: Completion by taking an examination

Continuous evaluation: none

Final evaluation: - written test

Evaluation: A: 100 – 95%; B: 94 – 89%; C: 88 – 83%; D: 82 – 77%; E: 76 – 71%; FX: 70% and less
Finished: exam - written test

Learning outcomes:
VV1 The student will know the basics of genetics and molecular basis of genetically determined pathological conditions
VV2 The student will understand genetic terminology, the mechanism of chromosomal aberrations, their consequences and the methodology of their visualization
VV3 The student will know the basics of mendelian and mitochondrial inheritance and the most common pathological conditions for each type of inheritance.
VV4 The student will understand the mechanism of cell cycle regulation as well as the causes and consequences of its failure
VV5 The student will learn the methodologies of recombinant DNA techniques and the possibilities of their practical application in diagnostics and therapy
VV6 The student will understand the nature of DNA polymorphisms and mutations and their relationship to selected pathological conditions.

Schedule of subject:
Lectures:
1) Meiosis and its specifics, mechanisms of chromosomal aberrations
2) Types of chromosomal aberrations and factors that influence their occurrence
3) Classical and molecular cytogenetics
4) Deregulation of the cell cycle and basics of carcinogenesis, epigenetics
5) Molecular basis of the most common forms of cancer and hereditary forms of cancer.
6) Fundamentals of recombinant DNA techniques, vectors and their applications, gene therapy, CRISPR/CAS9
7) DNA polymorphisms and their laboratory diagnosis (VNTR, SNP...)
8) DNA mutations as a cause of some human pathologies
9) Human genome and its characteristics (DNA types, their origin and significance - satellite DNA, mobile genetic elements, gene families)
10) Introduction to human genetics, history and basic genetic concepts and methods.
11) Monogenically determined normal and pathological traits autosomal inherited and traits linked to the X chromosome
12) Polygenic and multifactorial inheritance, mitochondrial inheritance. Fundamentals of population genetics

Practise:
1) Cell cycle - A) Preparation of smear stained slides of onion (Allium cepa) roots for microscopic observation of different stages of mitosis, calculation of mitotic index. B). Observation of onion roots after colchicine (or its derivatives)
2) C. elegans - model organism in genetics - Observation and identification of growth stages of C. elegans , determination of sex ratio (hermaphrodites vs males).
3) C. elegans - model organism in genetics, RNA interference - Induction and observation of RNA interference in the model organism C. elegans.
4) Analysis of chromosome number and structure from blood cells - Preparation of chromosome preparations
5) Analysis of chromosome number and structure from blood cells - Visualization of chromosomes - G banding (classical staining)
6) Molecular analysis - Isolation of genomic DNA followed by fluorescence detection and electrophoretic separation of nucleic acids by gel migration
7) Molecular analysis - PCR method - principle+detection of mutation of gene encoding transpeptidase in prokaryotic organisms;
8) Molecular Analysis - Visualization of chromosomes by hybridization method (FISH) - computer animation, human chromosome distribution, healthy human karyotype and chromosome aberrations - microscopic observation and animation
9) Mendelian concept of inheritance - Mendelian concept of inheritance
10) Heredity and sex
11) Population genetics
12) Application of the PCR method (PCR-RLFP) to determine the pathological consequences of coding sequence polymorphisms in humans (example: sickle cell anemia) - computer animation

Recommended reading:
1) NUSSBAUM, R. L. a spol.: Klinická genetika, 6. vyd., Triton, Praha, 2004, ISBN 8072544756.
2) PASSARGE, E.: Barevný atlas genetiky. Grada, Praha, 2019, ISBN 9788024730998.
3) PRITCHARD, D.J.-KORF, B.R.: Základy lékařské genetiky. Galén, Praha, 2007, ISBN 9788074925139.
4) SRŠEŇ,Š.-SRŠŇOVÁ, K.: Základy klinickej genetiky. Osveta, Martin, 2000, ISBN 8080631859.
5) STRACHAN,T.-GOODSHIP, J.-CHINNERY, P.: Genetics and Genomics in Medicine, Garland Science, Taylor and Francis Group, LLC, NY, 2015, ISBN 978-0-8153-4480-3.

Language requirements: Slovak

Notes:
The daily combined method consists of: lectures /on-line, distance, self-study/; practise /attend/
Self-study is supported by e-learning.
100% participation is required.

Prerequisite:
Biology I. (LMbD04)
Biology II. (LMbD11)

Course evaluation:
Assessed students in total: 755

A	B	C	D	E	FX
8%	11%	16%	21%	15%	28%

Lecturers:
Mgr. Patrícia Hockicková, PhD., examiner, instructor
prof. RNDr. Marie Korabečná, Ph.D., lecturer, examiner, instructor

Date of last change: 01.09.2026

Approved by: doc. Ing. Stanislava Blažíčková, PhD., MPH, univ.prof.