HUMAN GENETICS

MORPHOLOGY2

FIRST YEAR

2024/2025.

Course title:

HUMAN GENETICS

ECTS Credits: 6. Number of active classes per week: 4 (lectures: 2; practical classes: 2)

Teachers:

	Name and surname	Email	title
1.	Biljana Ljujic	bljujic74@gmail.com	Full Professor
2.	Olivera Milosevic-Djordjevic	olivera@kg.ac.rs	Full Professor
3.	Vladislav Volarevic	drvolarevic@yahoo.com	Full Professor
4.	Danijela Todorovic	dtodorovic@medf.kg.ac.rs	Associate Professor
5.	Marina Gazdic Jankovic	marinagazdic87@gmail.com	Associate Professor
6.	Danijela Cvetkovic	c_danijela@yahoo.com	Assistant Professor
7.	Nikolina Kastratovic	n_kastratovic@outlook.com	Teaching Assistant
8.	Dragana Papic	drmiloradovic7@gmail.com	Teaching Assistant
9.	Dragica Pavlovic	dragica.miloradovic8@gmail.c	Teaching Assistant

Course structure:

Module	Module title	Week	Theoretical classes per week	Practical classes per week	Teacher in charge:
1	Organization of the human genome	5	2	2	Assoc. prof. Marina Gazdić Janković
2	Significance of gene mutation and genetic determination of human traits	6	2	2	Full professor Vladislav Volarević
3	Developmental genetics and population genetics	4	2	2	Assoc. prof. Marina Gazdić Janković
					Σ 30+30=60

Examination Methods:

By fulfilling the pre-exam obligations and taking the written/test exam, the student can achieve a maximum of 100 points.

The grade is equivalent to the number of points earned (see tables). Points are earned in two ways:

ACTIVITY DURING THE CLASSES:

In this way, the student earns up to 30 points, by answering 2 questions from that week's classes at the practical classes and, in accordance with the demonstrated knowledge, gaining from 0 - 2 points.

FINAL EXAM:

The student takes the final test during the exam period. The test includes 35 questions. Each question is worth 2 points. In this way, the student can acquire 70 points, according to the attached grading scheme.

Determination of final grade:

To pass the exam, the student must earn the minimum of 51 total points and to fulfill the following:
1. to earn more than 50% points on activity during classes
2. to earn more than 50% points on the final exam, which includes total teaching material.

Grading system

Final grade	Total number of points Points grade	Description
10	91 – 100	Excellent
9	81 – 90	Exceptionally good
8	71 – 80	Very good
7	61 – 70	Good
6	51 – 60	Passing
5	< 51	Falling

LITERATURE:

textbook	authors	publisher	library	reading room
Emery's elements of medical genetics	Peter D. Turnpenny and Sian Ellard	Elsevier. 15th edition. 2017.	yes	yes

STUDY PROGRAM

THE MODULE 1: ORGANISATION OF HUMAN GENOME

Course unit 1 (1STWEEK):

EUKARYOTIC CHROMOSOMES

lecture 1 class	practice 1 class
Chemical composition of eukaryotic chromosome. DNA packaging Morphological features of chromosomes	Chemical composition of eukaryotic chromosome DNA packaging Morphological features of chromosomes Human karyotype Standardisation in human cytogenetics
lecture 1 class	practice 1 class
Human karyotype Standardisation in human cytogenetics	Student activity assessment

Course unit 2 (2ndWEEK):

NUCLEAR AND MITOCHONDRIAL GENOMES

lecture 1 class	practice 1 class
The human nuclear genome: structure and organisation Eukaryotic gene structure Number and length of human nuclear genes Gene polymorphism	Human nuclear and mitochondrial genome Eukaryotic gene structure Number and length of human nuclear genes Gene polymorphism

lecture 1 class	practice 1 class
The mitochondrial genome: structure and organisation Genes on the mitochondrial DNA Maternal inheritance	Student activity assessment

Course unit 3 (3st WEEK):

GENETIC RECOMBINATION

lecture 1 class	practice 1 class
Genetic recombination. Recombination in viruses. Conjugation, transformation and transduction Genetic recombination in bacteria	Genetic linkage The mechanism of crossing-over Gene mapping- practice problems
lecture 1 class	practice 1 class
Genetic recombination in eukaryotes - crossing-over-mechanism, modification of crossing over frequency Crossing over and genetic mapping Interference and coincidence	Student activity assessment

Course unit 4 (4th WEEK):

GENETIC ENGINEERING - RECOMBINANT DNA TECHNOLOGY

lecture 1 class	practice 1 class
Clone and cloning Gene cloning -methods of recombinant DNA technology	Gene cloning - methods of recombinant DNA technology. Reproductive cloning. Therapeutic cloning. Recombinant DNA in medicine
lecture 1 class	practice 1 class
Reproductive cloning Therapeutic cloning Recombinant DNA in medicine	Student activity assessment

Course unit 5 (5th WEEK):

EPIGENETICS. STEM CELLS AND THEIR APPLICATIONS.

lecture 1 class	practice 1 class
Introduction to epigenetics Biology of stem cells Types of stem cells Molecular mechanisms of pluripotency and reprogramming	Introduction to epigenetics Biology of stem cells Types of stem cells Molecular mechanisms of pluripotency and reprogramming Stem cells applications in regenerative medicine
lecture 1 class	practice 1 class
Stem cells applications in regenerative medicine	Student activity assessment

THE MODULE 2: THE SIGNIFICANCE OF GENE MUTATION AND GENETIC DETERMINATION OF HUMAN TRAITS

Course unit 6 (6th WEEK):

NUMERICAL CHROMOSOME ABERRATIONS

lecture 1 class	practice 1 class
Definition and mechanism of polyploidy	Polyploidy andaneuploidy Practice problems
lecture 1class	practice 1 class
Definition and mechanism of aneuploidy Types of aneuploidy Mixoploidy and chimerism	Student activity assessment

Course unit 7 (7th WEEK):

NUMERICAL AND STRUCTURAL CHROMOSOME ABERRATIONS

lecture 1 class	practice 1 class
Types and mechanism of chromosomal deletion Mechanism of chromosomal duplications Isochromosome and dicentric chromosome Types and mechanism of chromosomal inversion	Structural chromosome aberrations -deletion, duplications, inversion and translocations. Practice problems
lecture 1 class	practice 1 class
Types and mechanism of chromosomal translocations Differences between reciprocal translocations, Robertsonian translocations and insertions	Student activity assessment

Course unit 8 (8thWEEK):

CLINICAL FINDINGS IN CHROMOSOME ABERRATIONS

lecture 1 class	practice 1 class
Sex chromosome aneuploidies: Turner syndrome, Klinefelter syndrome, Triple X syndrome and XYY syndrome Autosomal aneuploidies: Down's, Edwards' and Patau's syndromes	Clinical findings in chromosome aberrations.
lecture 1 class	practice 1 class
Chromosome deletion: Cri-Du-Chat syndrome, Wolf-Hirschhorn syndrome, malignant diseases. Turner syndrome caused by X chromosome deletion Syndromes caused by chromosomal translocations Chromosomal aberrations-the cause of spontaneous abortions	Student activity assessment

Course unit 9 (9thWEEK):

GENE MUTATIONS

lecture 1 class	practice 1 class
Gene mutation: definition and types (somatic and germline mutations, spontaneous and induced mutations, micro and macro mutations) Point mutation – substitution (missense, nonsense, silent and neutral mutations), frameshift mutations (insertions and deletions) Dynamic mutation Spontaneous mutation rates	Mechanisms of gene mutation

lecture 1 class	practice 1 class
DNA repair DNA repair-deficiency disorder Mutagens	Student activity assessment

Course unit 10 (10thWEEK):

PATTERNS OF INHERITANCE

lecture 1 class	practice 1 class
Dominance and recessiveness Autosomal-dominant inheritance Autosomal recessive inheritance X-linked inheritance, Y-linked inheritance	Monohybrid inheritance Dihybrid inheritance Polygenic inheritance Practice problems- making and analysis of genealogical trees
lecture 1 class	practice 1 class
Sex limited inheritance Polygenic and multifactorial inheritance Genetic linkage Maternal inheritance	Student activity assessment

Course unit 11 (11th week):

PRENATAL DIAGNOSTIC OF CHROMOSOMOPATHY AND GENOPATHY

lecture 1class	practice 1 class
Prenatal diagnosis- indications Methods for non-invasive and invasive prenatal diagnosis Methods for invasive prenatal diagnosis: amniocentesis, chorionic villus sampling, cordocentesis. Preimplantation genetic diagnosis	Prenatal diagnosis of chromosomopathies and genopathy Methods in molecular genetics: hybridization, electrophoresis, blotting, PCR.
lecture 1class	practice 1 class
Prenatal diagnosis of genopathy Basic methods in molecular genetics: nucleic acid isolation, electrophoresis, PCR, hybridization tests	Student activity assesment

THE MODULE 3: DEVELOPMENTAL GENETICS AND POPULATION GENETICS

Course unit 12 (12th week):

SEX- DETERMINATIONAND DIFFERENTIATION

lecture 1class	practice 1 class
A Barr body- X- hromosom inactivation in females Role of X and Y chromosome sex diferentiation Autosomal chromosome genes responsible for gonad differentiation-SOX9, SF1, WT1	X- hromosom inactivation. Mary Lyon hypothesis Solving problem tasks.
lecture 1class	practice 1 class
Human sexual disorders. Sex reversions. Hermaphroditism.	Student activity assesment

Course unit 13 (13th week):

THE GENETICS OF IMMUNITY

lecture 1class	practice 1 class
Antigen Antibody structure and function Immune response mechanism Antibody genes HLA system	Multiple allelism Hierarchical relationship between alleles Codominant relationship between alleles Blood types ABO blood group system MN blood group system Rh blood group system Solving problem tasks.
lecture 1class	practice 1 class
Immunogenetics of blood groups: ABO blood group system MN blood group system Rh blood group system	Student activity assesment

Course unit 14 (14th week):

ONCOGENETICS. GENETICS OF AGING

lecture 1class	practice 1 class
Characteristics of the malignant cell Types of cancers according to the type of cell from which they arise. Carcinogens Chromosomal aberrations in cancer Viral origin of cancer	The genetic basis of cancer – chromosomal aberrations and gene mutations in malignancies.
Practice 1 class	practice 1 class
Genetic basis of cancer: protooncogene, oncogene, cancer-suppressor gene p53 gene Aging	Student activity assesment

Course unit 15 (15th week):

POPULATION GENETICS

lecture 1class	practice 1 class
Definition and characteristics of human populations. Genetic structure of a population- The Hardy-Weinberg equilibrium principle. Panmixia.	Genetic structure of a population-The Hardy-Weinberg equilibrium principle. Solving problem tasks.
lecture 1class	practice 1 class
Factors that disrupt the population's genetic structure: natural selection, mutations, migrations, genetic coincidence Genetic load- consanguineous marriages.	Student activity assesment

Schedule of teaching lectures

INTERNAL CLINIC HALL

WEDNESDAY

13:35 - 15:05

Schedule of practical classes

THURSDAY	
R9-1	**R9-2**
08:00 - 09:30 **I group** **09:30 – 11:00** **II group** **11:00 - 12:30** **III group** **12:30 – 14:00** **IV group**	**08:00 - 09:30** **V group** **09:30 – 11:00** **VI group** **11:00 - 12:30** **VII group**

SCHEDULE

module	week	type	title of the lecture	teacher
1	1	**L**	Chromosomes of eukaryotes. Human karyotype.	Assoc. prof. Marina Gazdić Janković
1	1	**P**	Chromosomes of eukaryotes. Human karyotype.	Dragana Papić Dragica Pavlović Nikolina Kastratović
1	2	**L**	Nuclear and mitochondrial genome	Prof. Vladislav Volarević
1	2	**P**	Nuclear and mitochondrial genome	Dragana Papić Dragica Pavlović Nikolina Kastratović
1	3	**L**	Genetic recombination	Prof. Vladislav Volarević
1	3	**P**	Genetic recombination	Dragana Papić Dragica Pavlović Nikolina Kastratović
1	4	**L**	Genetic engineering - recombinant DNA technology	Prof. Vladislav Volarević
1	4	**P**	Genetic engineering – recombinant DNA technology	Dragana Papić Dragica Pavlović Nikolina Kastratović
1	5	**L**	Epigenetics. Stem cells and their applications.	Assoc. prof. Marina Gazdic Jankovic
1	5	**P**	Epigenetics. Stem cells and their applications.	Dragana Papić Dragica Pavlović Nikolina Kastratović
2	6	**L**	Numerical chromosome aberrations	Prof. Vladislav Volarević
2	6	**P**	Numerical chromosome aberrations	Dragana Papić Dragica Pavlović Nikolina Kastratović

SCHEDULE

module	week	type	title of the lecture	teacher
2	7	**L**	Numerical and structural chromosome aberrations	Prof. Vladislav Volarević
2	7	**P**	Numerical and structural chromosome aberrations	Dragana Papić Dragica Pavlović Nikolina Kastratović
2	8	**L**	Clinical findings in chromosome aberrations	Assoc. prof. Marina Gazdić Janković
2	8	**P**	Clinical findings in chromosome aberrations	Dragana Papić Dragica Pavlović Nikolina Kastratović
2	9	**L**	Gene mutations	Prof. Vladislav Volarević
2	9	**P**	Gene mutations	Dragana Papić Dragica Pavlović Nikolina Kastratović
2	10	**L**	Patterns of inheritance	Assoc. prof. Marina Gazdić Janković
2	10	**P**	Patterns of inheritance	Dragana Papić Dragica Pavlović Nikolina Kastratović
2	11	**L**	Prenatal diagnostic of chromosomopathy and genopathy	Assoc. prof. Marina Gazdić Janković
2	11	**P**	Prenatal diagnostic of chromosomopathy and genopathy	Dragana Papić Dragica Pavlović Nikolina Kastratović

SHEDULE

module	week	type	title of the lecture	teacher
3	12	**L**	Sex- determination and differentiation	Assoc. prof. Marina Gazdić Janković
3	12	**P**	Sex- determination and differentiation.	Dragana Papić Dragica Pavlović Nikolina Kastratović
3	13	**L**	The genetics of immunity	Assoc. prof. Marina Gazdić Janković
3	13	**P**	The genetics of immunity	Dragana Papić Dragica Pavlović Nikolina Kastratović
3	14	**L**	Oncogenetics	Assoc. prof. Marina Gazdić Janković
3	14	**P**	Oncogenetics	Dragana Papić Dragica Pavlović Nikolina Kastratović
3	15	**L**	Population genetics	Prof. Vladislav Volarević
3	15	**P**	Population genetics	Dragana Papić Dragica Pavlović Nikolina Kastratović