HUMAN GENETICS	
GENERAL INFORMATIONS	
Course coordinator	Prof. Jasenka Wagner Kostadinović, MMedBiochem, PhD
Assistant/Associate	Prof. Marija Heffer, MD, PhD Asst. Prof. Vedrana Ivić, MEdBiol, PhD
Study Programme	Undergraduate university study of Medical laboratory diagnostics
Status of the course	manatory
Year of study, semester	2nd year, 4th semester
ECTS	**3**
Workload (hours)	Lectures: 15 ; Seminars:5; Laboratory exercises:15
Expected number of students	30-35
COURSE DESCRIPTION	
Course objectives	
Human genetics is one of the most rapidly developing fields in medicine, while molecular genetics is integrated into all parts of medical science. Every medical practitioner of the 21st century, in the post-genomic age, will have to have a deep and extensive knowledge of the basic principles of human genetics and their wide application in health and disease. A human genetics course should provide the knowledge and skills that make this possible.	
Enrolment requirements and entry competencies	
Passed all exams of previous years of study	
Learning outcomes at the Programme level	
1.1., 1.2., 2.1, 2.2., 2.7, 3.1, 3.2	
Learning outcomes	
After completing lectures, seminars and exercises, independent study and passing the exam, students will be able to: 1. determine the type and mode of inheritance of the disease 2. choose and implement appropriate laboratory tests to diagnose genetic diseases 3. interpret the findings with regard to the etiology of the disease and the technical aspects of the laboratory test 4. predict the risk of the disease in the offspring and the form of disease progression in the patient 5. know and apply the ethical norms of the profession and the principles of genetic counseling	
Course content	
Lectures: Genotype and phenotype. DNA as a hereditary material. Regulation of gene expression. Somatic and germline mutations. Meiosis, gametogenesis, fertilization. Medel's laws of inheritance. Family tree. Multiple alleles and complex traits. Anticipation. Mosaicism. Uniparental disomy. Genomic imprinting. Mitochondrial inheritance. Monogenic diseases. Disease models with multifactorial inheritance. Genomic imprinting. Uniparental disomy. Prader-Willie and Angelman syndrome. Epigenetics and tumor growth. Allele frequency in the population. Segregation analysis. Genetic linkage analysis. Medical and social interventions. Distribution of diseases with polygenic inheritance. Gene polymorphism. Techniques for identification of genes causing multifactorial diseases. Distinguishing genetic factors from environmental influences in tumor development. Oncogenes. Tumor suppressor genes. Epigenetics and cancer. Genetics of the most common cancers. Genetic counseling for families with hereditary tumors. Immunity. Early signs of hereditary diseases that appear at an early, pubertal and young age: autism, schizophrenia, gender dysmorphism. Screening of at-risk populations and at-risk individuals.	

Epigenetics and brain development. Mapping of regions in identical twins, brothers and unrelated individuals. Biochemical genetics.

Seminars: Screening of the high-risk population. Testing of carriers of autosomal recessive and sex-linked traits. Presymptomatic diagnosis of autosomal dominant diseases. Registers of genetic diseases. Types and mechanisms of genetic predisposition. Ways to detect genetic predisposition to the development of common diseases. Y chromosome microdeletions. Causes of female infertility. Assisted conception. Implications for genetic diseases. Preimplantation diagnostics. Risk calculation. Ethical dilemmas in the context of the wider community. Examples from clinical practice. Techniques used in prenatal diagnosis. Prenatal screening. Indications for prenatal diagnostics. Non-invasive prenatal diagnostics. Prenatal therapy. Fertilization and implantation. Fertility problems.

Exercises: PCR, RT-PCR, Southern blot. Practical resolution of molecular analysis findings. Principles of drawing a family tree. Examples of tasks with monogenic autosomal dominant and autosomal recessive diseases. Disease symptoms characteristic of hereditary malformations. Indications for karyogram analysis, method of sample collection and transport, preparation of chromosome slides, staining techniques and chromosome analysis. Karyograms of characteristic chromosomal abnormalities. Disorders of the number of sex chromosomes. Chromosomal disorders and behavioral changes as a phenotypic marker. Disorders of sexual differentiation. Chromosome breakage syndromes. Indications for karyogram analysis. In situ genetic analysis techniques: FISH. Chromosome staining. Comparative genomics.

Mode of teaching

Lectures, Problem solving seminars, Laboratory exercises

Student obligations

Attendance at all forms of classes is mandatory and student must be prepared for seminars and exercises. Absence from the seminars and exercises must be compensated. It is forbidden to use a mobile phone during classes and exams.

Monitoring student work *(Connectivity of learning outcomes, teaching methods and grading)*

Exam method: written and oral exam.

Teaching activity	ECTS	Learning outcome	Student activity	Assessment methods	Grade points	
					Min.	Max.
Attending classes	1	1-5	Class attendance	Attendance record	8	16
Seminars		1-5	Preparation of seminar	Seminar presentation		
Exercises		1-5	entrance exams, performing exercises, keeping work diary	work diary, entrance exam		
Final exam	2	1-5	Studying for the final exam	Written exam	26	42
				Oral exam	26	42
Total	**3**				**60**	**100**

Evaluation of the final written exam:

Percentage of correct answers (%)	Ocjenski bodovi
60,00-64,99	26
65-69,99	28
70,00-74,99	30
75-79,99	32
80-84,99	34
85-89,99	36
90,00-94,99	39
95-100	42

Calculation of final grade:

Grade points earned in the final exam are added to the grade points earned during the course. Grading in the ECTS system is done by absolute distribution, i.e. based on total achievement and is compared to the numerical system in the following manner: A - excellent (5): 90-100 grade points; B - very good (4): 80-89.99 grade points; C - good (3): 65-79.99 grade points; D - sufficient (2): 50-64.99 grade points.

Required reading (available in the library and through other media)

Title	Number of copies in the library	Availability through other media
Turnpenny P, Ellard S: Emeryeve OSNOVE MEDICINSKE GENETIKE, 14th Ed., Medicinska naklada, Zagreb, 2011.	10	
Emery's elements of medical genetics, 15th edition, 2017.		online

Additional reading

The latest scientific and professional literature recommended by professors.

Course evaluation procedures

Anonymous, quantitative, standardised student survey on the course and the teacher's work implemented by the Quality improvement office of the Faculty of Medicine Osijek.

Note /Other

E-learning is not included in the class quota, but it is used in teaching and it contains links to various sites and video and audio materials available on websites.