MEDICAL GENETICS	
GENERAL INFORMATION	
Course coordinator	Associate Professor Jasenka Wagner Kostadinović, PhD
Assistant/Associate	Professor Marija Heffer, MD, PhD Professor Borut Peterlin, PhD Associate Professor Silvija Pušeljić, MD, PhD
Study Programme	Integrated undergraduate and graduate university study of Medicine
Status of the course	Mandatory
Year of study, semester	6th year, 11th semester
ECTS	**4**
Workload (hours)	Lectures (27); Seminars (18)
Expected number of students	70
COURSE DESCRIPTION	
Course objectives	
To identify a pathophysiological state that has a genetic etiology. Properly select and interpret the result of a genetic test, understand the consequences of genetic and epigenetic changes on health, interpret the family tree and assess the risk of genetic disease in other family members.	
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, 2.3., 3.2., 3.4.,3.5., 4.2.	
Learning outcomes (5-10)	
After listening to lectures and seminars, independent learning and passing the exam, students will know how to: 1. determine the type and mode of inheriting the disease 2. select appropriate laboratory tests to diagnose genetic disease 3. Interpret the obtained results of genetic testing with regard to the etiology of the disease 4. predict the risk for disease in the offspring and the level of disease progression in the patient 5. know the ethical principles of the profession and the principles of genetic counseling of the patient and his family members	
Course content	
Lectures Organization of hereditary matter: structure and function of DNA and RNA, organization of chromatin, human karyogram. Chromosomopathies: numerical and structural aberrations, chromosomal syndromes, microdeletion syndromes, clinical molecular cytogenetics. Inheritance patterns: Mendel's and non-Mendel's inheritance (mitochondrial, uniparent disomy, genomic imprinting, dynamic mutations, mosaicism, digenic mode of inheritance); polygenic and multifactorial inheritance. Monogenic diseases. Polygenic diseases. Oncogenetics: genetic basis of malignant transformation, proto-oncogenes and mechanisms of their activation, tumor suppressor genes, cell cycle control disorders, tumorigenesis.	

Population genetics: characteristics of population in equilibrium, Hardy-Weinberg principle, factors that affect genetic balance, genetic polymorphisms and the importance of population-genetic research in medicine.
Developmental genetics: gametogenesis, fertilization, early embryonic development, sex determination, growth and differentiation, differential gene activity, differentiation of teratogenic and hereditary factors.
Congenital anomalies and dysmorphic syndromes. Reproductive genetics (assisted reproduction techniques, genetic causes of infertility). Prenatal diagnostics: invasive methods (chorionic villus biopsy, amniocentesis, cordocentesis) and non-invasive methods (ultrasound and biochemical screening, analysis of free fetal DNA from maternal blood), preimplantation diagnostics.
Genetic counseling: making a clinical diagnosis, choosing a diagnostic method, calculating risk, providing genetic information in accordance with the basic ethical principles and methods of genetic counseling. Specific problems in genetic counseling (non-penetrance, variable expression, phenocopies, genetic heterogeneity, consanguinity, false paternity, incest). Screening tests for genetic diseases (population, familial, neonatal, prenatal).
Immunogenetics (primary inherited immune disorders, secondary immunodeficiencies).
Neurogenetics. Pharmacogenomics, treatment of genetic diseases, gene therapy. Cardiac genetics.
Biochemical genetics. Epigenetics.

Seminars

Molecular biology techniques: Southern and Northern blot, restriction fragment length polymorphism (RFLP), polymerase chain reaction (PCR), reverse transcription (RT), real time polymerase chain reaction (real time PCR), Sanger sequencing, next generation sequencing (NGS), genome / exome / clinical exome sequencing; method selection, interpretation of laboratory analysis results.
Cytogenetic techniques: classical (G- and C-banding) and molecular cytogenetics techniques (fluorescent in situ hybridization (FISH) and comparative genomic hybridization on a microarray (aCGH); method selection, interpretation of laboratory test results.
Ethics in medical genetics. Genetic resources on the Internet: online genomic databases, web pages with information on expert centers, clinical studies, accredited genetic laboratories, orphan drugs. Interpretation of family genetic background and identification of genetic problems.
Genetics of mental retardation. Mitochondrial and metabolic diseases. Genetic counseling-role-playing.

Mode of teaching

Lectures; Seminars

Student obligations

Attendance at all forms of classes is mandatory, and the student must access all knowledge tests. A student may justifiably miss 30% of each form of instruction. Absence from the seminars must be compensated.

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

Teaching activity	ECTS	Learning outcome	Student activity	Assessment methods	Grade points	
					Min.	Max.
Lectures	0,5	1-5	Class attendance	Attendance records	5	10
Seminars	0,5	1-5	Attendance and active participation in seminars	Attendance records, presentations, written essay	10	20
Written and oral exam	3,0	1-5	Learning for written and oral exam	Written and oral exam	35	70
Total	**4**				**50**	**100**

Calculation of final grade:

Grades achieved in the written and in the oral exam are added to the grade points achieved during the course. The evaluation is performed by absolute distribution, i.e. on the basis of the final achievement and is compared with the numerical system as follows:

A - excellent (5): 80-100 points; B - very good (4): 70-79.99 points; C - good (3): 60-69.99 points; D - sufficient (2): 50-59.99 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, 14. izdanje, Medicinska naklada, Zagreb, 2011.	11	

Additional reading

1. Emery's elements of medical genetics, 15th edition, 2017. (available online)
2. The latest scientific and professional literature recommended by professors

Course evaluation procedures

Anonymous, quantitative, standardized student survey on the subject and work of teachers conducted by the Office for Quality of the Medical Faculty Osijek.

Note /Other

E-learning is not included in the norm of subject hours, but is used in teaching and contains links to various pages, video and audio materials available on the website.