

GENERAL INFORMATION		
Course name	Human Genetics	
Course director	Prof. Jasenka Wagner Kostadinović, PhD	
Assistants	Prof. Marija Heffer, MD, PhD Prof. Feodora Stipoljev, PhD Prof. Borut Peterlin, PhD	
Study program	Integrated undergraduate and graduate university study program Medical Studies in German	
Course status	Mandatory	
Year of study, semester	6 th year, 12 th semester	
Credits allocated and form of instruction	ECTS student workload	3
	Number of teaching hours (L+S+E)	40 (20+10+10)
COURSE DESCRIPTION		
Course objectives		
Learning the basics of genetics and diagnostic methods in genetics. Familiarizing students with epidemiology, genetic basis, pathophysiology and clinical picture of the most common hereditary diseases.		
Course requirements		
Completed courses: Biology, Biochemistry, Pediatrics, Gynecology, Internal medicine, Oncology		
Learning outcomes at the Programme level		
1.1., 1.2., 2.1., 2.3., 3.2., 3.4., 3.5., 4.2.		
Expected learning outcomes (5-10 learning outcomes)		
<p>KNOWLEDGE</p> <ol style="list-style-type: none"> Determine which changes in DNA sequences lead to disease Distinguish between phenotype and genotype and explain them Explain terms: nonpenetrance, pleiotropy, variable expressivity, anticipation, mosaicism Explain the terms of uniparental disomy and genomic imprinting (level of a gene, chromosome, genome) List hereditary trinucleotide repeat diseases Explain genome instability Be familiarized with the national strategy for reducing risk factors regarding the occurrence of polygenic diseases Know which screening tests exist in the Republic of Croatia and when they are used Know how to discover a genetic predisposition for developing common diseases Determine personal risk and predilective factor Explain haplotype and gene polymorphism Know the most common monogenic diseases, their causes and modes of inheritance Determine methods for identifying the genes causing multifactorial diseases Differentiation of genetic factors from environmental impact in tumor development List inherited immunodeficiencies Explain how genetics affects drug metabolism Identify the type and mode of inheriting a disease Select a laboratory test Understand the molecular basis of the pathophysiology of a disease Explain chromosomopathy and its most common types Independently and critically interpret scientific literature in the field of medical genetics Explain the genetic basis of malignant transformations List and explain the genetic diseases compatible with life, if treated 		

24. List hereditary diseases with late onset

SKILLS

1. Independently identify a genetic problem
2. Classify genetic problems for genetic counseling
3. Independently select and interpret tests on suspicion of genetic causes of infertility
4. Calculate risk of the occurrence of disease (monogenic, polygenic diseases, Bayes' theorem)
5. Interpret laboratory results (molecular-genetic, molecular-cytogenetic, cytogenetic)
6. Identify and be able to draw a family tree
7. Independently calculate allele frequencies in the population and apply the Hardy-Weinberg principle
8. Independently search online genetic databases
9. On the basis of a clinical image, diagnose a genetic disease
10. Relay the information to the patient in accordance with the basic principles of genetic counseling

Course content

Organization of hereditary material: structure and function of DNA and RNA, chromatin organization, human karyogram.

Genetic, chromosomal and genomic mutations: mechanism of spontaneous and induced mutations; physical, chemical and biological mutagens, numerical and structural aberrations, chromosomal syndromes.

Types of inheritance: Mendelian (autosomal and sex-linked) and non-Mendelian inheritance (mitochondrial, uniparental disomy, genomic imprinting, dynamic mutations and mosaicism); polygenic and multifactorial inheritance.

Molecular biology techniques: restriction endonucleases and vectors; cDNA and DNA libraries, hybridization probes, Southern and Northern blot, sequencing, polymerase chain reaction (PCR), DNA microchips, NGS.

Cytogenetic techniques: classical (G, R and C-bands for identification of chromosomes) and molecular cytogenetics techniques (FISH, mFISH, CGH, aCGH, microdissection and reverse chromosome painting).

Oncogenetics: genetic basis of malignant transformations, proto-oncogenes and mechanisms of activation, tumor-suppressor genes, cell cycle regulation disorders, tumorigenesis.

Population genetics: characteristics of the population in equilibrium, Hardy-Weinberg principle, factors altering the genetic balance, genetic polymorphism and the importance of population-genetic research in medicine.

Prenatal diagnostics: invasive and non-invasive methods, pre-implantation diagnostics.

Genetic counseling: clinical diagnosis, diagnostic method selection, risk calculation, relaying genetic information in accordance with basic ethical principles and methods of genetic counseling. Specific problems in genetic counseling (gene nonpenetrance, variable expression, phenocopies, genetic heterogeneity, consanguinity, false paternity).

Developmental genetics: gametogenesis, fertilization, early embryonic development, sex determination, growth and differentiation, differential gene activity, differentiation of teratogenic and hereditary factors, assisted reproduction methods.

Form of instruction	<input checked="" type="checkbox"/> lectures	and	<input checked="" type="checkbox"/> individual assignments
	<input checked="" type="checkbox"/> seminars		<input type="checkbox"/> multimedia and internet
	workshops		<input type="checkbox"/> laboratory
	<input checked="" type="checkbox"/> exercises		<input type="checkbox"/> mentoring activities
	<input type="checkbox"/> distance learning		<input type="checkbox"/> other
	<input type="checkbox"/> field course		

Student obligations

Come to class prepared by studying the recommended literature for each unit and actively participate in all forms of instruction. The student must participate in at least 70% of classes to pass the course.

Monitoring student learning

Attendance	x	Active participation	x	Seminar paper		Experimental work	x
Written exam	x	Oral exam	x	Essay		Research	
Project		Continuous assessment		Paper		Practical work	x
Portfolio							

Assessment and evaluation of students during class and on the final exam

Students' performance will be evaluated during class and on the final exam. Students are evaluated numerically and descriptively (insufficient (1), sufficient (2), good (3), very good (4), excellent (5)). During classes, a student can earn a maximum of 100 points. Students can earn a maximum of 20 points during classes through different types of activities. On the final exam, students can earn a maximum of 80 points. The final grade represents the sum of the points earned during classes and on the final exam.

Mandatory reading

1. Murken, Grimm, Holinski-Feder: Taschenlehrbuch humangenetik isbn: 9783132416871, 9. teilaktualisierte auflage 2017, thieme verlag

Additional reading

1. Read / Donnai: Angewandte Humangenetik, ISBN 9783110194654, DeGruyter Verlag, 1. Auflage 2008
2. Schaaf / Zschocke: Basiswissen Humangenetik, ISBN: 9783540712220, Springer Verlag, 2008
3. Peter Turnpenny and Sian Ellard. Emerys Elements Of Medical Genetics, 15th edition, Elsevier, 2017

The number of copies of mandatory reading in proportion to the number of students currently taking this course

<i>Title</i>	<i>Number of copies</i>	<i>Number of students</i>
1. Murken, Grimm, Holinski-Feder: Taschenlehrbuch humangenetik isbn: 9783132416871, 9. teilaktualisierte auflage 2017, thieme verlag	A purchased license for online textbooks shall be used https://bfdproxy48.bfd-online.de/login.htm?back=http%3a%2f%2fpartner.bfd-online.info.bfdproxy48.bfd-online.de%2fameos%2fbfdAboGateway%3fabold%3d264117 Access will be granted to all students enrolled in the study program	

Quality monitoring methods ensuring the acquisition of knowledge upon completion, skills and competences

The quality of course performance is monitored through an anonymous student survey on the quality of the organization and conduction of classes, the course content and the work of professors. The usefulness of the lectures from the students' perspective, the curriculum content, the professor preparedness, the clarity of the presentation, the amount of new content and the quality of the presentation are evaluated. The curriculum and its execution are administratively compared. The participation of students in lectures and exercises, as well as the excuses for missing classes, are controlled and analyzed.