

NAME OF THE COURSE		Medical Genetics				
Code	PMB727	Year of study	1			
Course teacher	Jelena Korac Prlic, PhD, Assistant Professor	Credits (ECTS)	3			
Associate teachers		Type of instruction (number of hours)	L	S	E	F
			10	15	5	
Status of the course	elective	Percentage of application of e-learning				
COURSE DESCRIPTION						
Course objectives	Aim of this course is to introduce students with basics of medical genetics with examples of genetic diseases. Students will be introduced to diagnostic technics and therapy in the field of medical genetics.					
Course enrolment requirements and entry competences required for the course	None					
Learning outcomes expected at the level of the course (4 to 10 learning outcomes)	<ul style="list-style-type: none"> Describe the structure of human genome and 'average' genes. Explain the definitions and learn basic rules of inheritance using basic examples. Learn how to use the genetic terminology. Significance of mutations. Explain the autosomal and sex-linked inheritance. Learn to recognize correct inheritance type. Understanding the genetic and environmental background of certain monogenic diseases, polygene diseases, chromosomal disorders. Examples. Knowledge of the method of prenatal genetic testing; Ethical and legal issues in medical genetics. Use of basic genetic techniques in the context of basic genetic discoveries. Basic examples of pharmacogenomics importance. Understanding the connection between cancer genetics and polygenic phenotypic characteristics. Learning the importance of modern genetic breakthroughs including gene therapy, genetically modified organisms and stem cell research. Comparison and usage of different gene and protein databases. - During the course, students learn how to communicate, present data and discuss about relevant scientific topics, and how to synthesize learned material. 					
Course content broken down in detail by weekly class schedule (syllabus)	<p>Lectures:</p> <ol style="list-style-type: none"> 1. Introduction to Medical genetics. Human genome project. (2 hours) 2. RNA genes. RNAi. Mutations and aberrations. Functional genomics & proteomics. (2 hours) 3. DNA analysis. (2 hours) 4. Patterns of Inheritance (2 hours) 5. Gene therapy. (2hours) <p>Seminars:</p> <ol style="list-style-type: none"> 1. Hemoglobin and the Hemoglobinopathies, Inborn Errors of Metabolism 2. Phamacogenetics. Single gene disorders. 3. Congenital malformations. Chromosome disorders 4. Cancer genetics. 					

	5. Genetic factors in common disorders. 6. Prenatal testing, Screening for genetic disease. Practicals: 1. Bioinformatics: Database search. DNA sequence analysis. OMIM. 2. Odds, probabilities. Risk calculation.					
Format of instruction	<input checked="" type="checkbox"/> lectures <input checked="" type="checkbox"/> seminars and workshops <input checked="" type="checkbox"/> exercises <input type="checkbox"/> <i>on line</i> in entirety <input type="checkbox"/> partial e-learning <input type="checkbox"/> field work			<input type="checkbox"/> independent assignments <input type="checkbox"/> multimedia <input type="checkbox"/> laboratory <input type="checkbox"/> work with mentor <input type="checkbox"/> (other)		
Student responsibilities	Attendance on lectures and seminars					
Screening student work (<i>name the proportion of ECTS credits for each activity so that the total number of ECTS credits is equal to the ECTS value of the course</i>)	Class attendance	1	Research		Practical training	
	Experimental work		Report		(Other)	
	Essay		Seminar essay		(Other)	
	Tests		Oral exam		(Other)	
	Written exam	2	Project		(Other)	
Grading and evaluating student work in class and at the final exam	Grading will be conducted based on activities in class, seminar work, practical exercises in the laboratory and final written exam.					
Required literature (available in the library and via other media)	Title			Number of copies in the library	Availability via other media	
	Emery's Elements of Medical Genetics – Turnpenny P and Ellard S., 15th ed. Elsevier, 2017.					
Optional literature (at the time of submission of study programme proposal)	Human molecular genetics. Strachan T, Read AP. 4th ed. New York (NY): Garland Science, Taylor & Francis Group; 2010.					
Quality assurance methods that ensure the acquisition of exit competences	Students' evaluation via anonymous questionnaires at the end of the course. The survey is conducted according to the rules of the University of Split.					
Other (as the proposer wishes to add)						