Name of the course	**Medical Genetics**			**Code**	
Type of study program Cycle	Integrated study program, medicine			**Year of study**	2nd
Credits (ECTS) :	**4**	*Semester*	II	Number of hours per semester (l+e+s)	45 (20+20+5)
Status of the course:	required	*Preconditions:*		*Comparative conditions:*	
Access to course:	second year students			*Hours of instructions*:	According to schedule

Course teacher:	Head: Prof. Katarina Vukojević
Consultations:	e-mail correspondence
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Associate teachers	Prof. Violeta Šoljić Senior assistant Una Glamočlija Senior assistant Maja Barbarić Assistant Martina Vukoja
Consultations:	e-mail correspondence
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The aims of the course:	The objectives of this course are: to introduce medical students with basic facts about medical genetics, introduce to the concepts of human medical genetics and appreciation of the genetic perspective on health and disease.
Learning outcomes (general and specific competences):	On completion of the course, the student should achieve general and specific outcomes. General outcomes: The course intends to give basic medical genetic knowledge about the structure and function of the human genome as well as the importance of relevant genetic factors for origin of diseases, abnormalities and developmental disorders in humans. Apply personal qualities of personality (team work and personal contribution, interest, active listening, and building positive relationships with members of the group). Specific outcomes: Demonstrating and understanding the structure of the human genome and function and know and understand basic concepts for the expression of most studied genes. Explain the definitions and learn basic rules of inheritance using basic examples. Know and have

	understanding for different genetic factors of importance for the origin of hereditary diseases and for the genetic variation of normal properties. Learn how to use the genetic language. Explain the significance of genetic mutations (the autosomal and sex-linked inheritance). Know and be able to use basic genetic concepts and identify Mendelian inheritance patterns. Describe, explain and outline principles of basic medical genetic techniques in the context of basic genetic achievements. Explain the basic concepts of pharmacogenomics importance. Describe and analyse the connection between cancer genetics and polygenetic phenotypic characteristics. Learning the importance of modern medical genetic and the scientific principles that are the foundation of current approaches to the diagnosis and treatments (stem cell therapies, gene therapy and genetically modified organisms). Describe, explain and outline principles of usage of different gene and protein databases. During the course, students learn how to communicate, present data and discus about relevant scientific topics, and how to synthesize learned material. Knowledge about medical genetics will be useful tool in recognizing, treating and preventing genetic disorders. Outcomes will be evaluated with continuous assessment, quizzes seminars and colloquium exercise and active forms of learning during exercises, lectures and seminars (quizzes for each unit), and the final practical, written and oral exam.
Course content (Syllabus):	Course consists of 10 units, assessment in seminars, and MCQ test. Each thematic unit includes: 2 hours of lectures, 2-3 hours of seminars and 0-1 hours of exercises. L1 (2 hours) – Introduction to Medical genetics L2 (2 hours) – Functional genomics and proteomics L3 (2 hours) – Genomics and the Human Genome Project L4 (2 hours) – Pharmacogenomics L5 (2 hours) – RNA genes and RNAi L6 (2 hours) – Mutations and aberrations L7 (2 hours) – DNA analysis L8 (2 hours) – Mitochondrial inheritance and human development L9 (2 hours) – Gene therapy. Genetically modified organisms (GMO) L10 (2 hours) – Epigenetics S1 (3 hours) – Chromosomes. DNA analysis techniques. S2 (3 hours) – Inheritance patterns (Mendelian and Non-Mendelian) and genetic counselling S3 (3 hours) – Applications to public health - screening and identification of populations at risk S4 (3 hours) – Carcinogenesis and common genetic factors S5 (3 hours) – Genes and molecular mechanisms underlying human disease

	S6 (3 hours) – Genetic background of congenital anomalies S7 (2 hours) – Gene ethics E1 (1 hour) – Introduction to Cytogenetics laboratory E2 (1 hour) – Primer design for genetic testing E3 (1 hour) – Bioinformatics (database search and OMIM) E4 (1 hour) – Cloning, transgenic animals, gene therapy E5 (1 hour) – Odds, probabilities, Bayes' theorem.			
Format of instruction (mark in bold)	**Lectures**	**Exercises**	**Seminars**	**Independent assignments**
	Consultations	Work with mentor	**Field work**	Other
	Remarks: The teaching of each unit begins with a lecture, followed by seminars and exercises. The course is based on self-study. Information about different activities such as assignments and submission dates are on the website of the course. Communication between students and teachers take place primarily via the website and via e-mail. It is a requirement that the participants have access to the Internet. At the seminars, students receive problem tasks that are solved in small groups, at the end of the seminar is a quiz-test, and then students discuss the correct answers with explanations of problems.			
Student responsibilities	Final exam; active participation in seminars; tasks; MCQ tests; attendance and participation in class. Students will be evaluated based on: • Active participation in seminars. • Preparation of teaching units for seminars • Reading of teaching texts and developing their own critical thinking about the material and express those views. • work in small groups			
Screening student work (mark in bold)	**Class attendance**	**Class participations**	**Seminar essay**	**Practical training**
	Oral exam	**Written exam**	**Continuous assessment**	Essay

Detailed evaluation within a *European system of points*

STUDENTS RESPONSIBILITIES	**HOURS**	**PROPORTIONS OF ECTS CREDITS**	**PROPORTIONS OF MARK**
Class attendance and participations	30	0,5	0%
Seminar essay	20	0,5	10%
Written exam	50	3,0	90%
Practical work	5	0	0%

Further clarification: Final written exam (90% of grade) Test points: 27-33 = (2); 33-39= (3); 40-45 = (4); 46-50 = (5); Reports from oral presentations during seminars (10% of grade) Final score: The final score is the sum of = complete written (90%) + oral presentations during seminars (10%).

Required literature:	Emery's Elements of Medical Genetics – Peter D Turnpenny, Sian Ellard, 14th edition, Elsevier, 2012.
Optional literature:	Essential Medical genetics – Tobias E.S, Connor M, Ferguson-Smith M, 6th edition, Wiley-Blackwell, 2011
Additional information about the course	Students responsibilities are in accordance to Rules of studying and Deontological code of MEFMO students. Methods of monitoring the quality of teaching: student survey Quality control analysis by the students and teachers Analysis of passing the exams The report of the Office for the quality of teaching

Annexes: calendar classes

The number of teaching units	TOPICS AND LITERATURE
I.	Title: Introduction to Medical genetics
	Short description: Basic principles of Medical genetics; mitosis, meiosis and chromosomes
	Literature: required and optional
II.	Title: Functional genomics and proteomics
	Short description: Genome structure, genetic mapping, basic principles of proteomics
	Literature: required and optional
III.	Title: Genomics and the Human Genome Project
	Short description: Determining the sequence of nucleotide base pairs that make up human DNA, and of identifying and mapping all of the genes of the human genome from both a physical and a functional standpoint.
	Literature: required and optional
IV.	Title: Pharmacogenomics

	Short description: The role of the genome in drug response. Its name (pharmaco + genomics) reflects its combining of pharmacology and genomics
	Literature: required and optional
V.	Title: RNA genes and RNAi
	Short description: Description of biological process in which RNA molecules inhibit gene expression or translation, by neutralizing targeted mRNA molecules.
	Literature: required and optional
VI.	Title: Mutations and aberrations
	Short description: Description of a missing, extra, or irregular portion of chromosomal DNA, gene mutations and aberrations
	Literature: required and optional
VII.	Title: DNA analysis
	Short description: DNA profiling to determine an individual's DNA characteristics
	Literature: required and optional
VIII.	Title: Mitochondrial inheritance and human development
	Short description: The DNA of cytoplasmic organelles is inherited in a non-Mendelian manner. This pattern of inheritance is generally referred to "maternal inheritance." Implications to human development
	Literature: required and optional
IX.	Title: Gene therapy. Genetically modified organisms (GMO)
	Short description: Utilisation of different vectors to deliver genes which can cure disease in humans. Implications of gene therapy
	Literature: required and optional
X.	Title: Epigenetics
	Short description: The study of changes in organisms caused by modification of gene expression rather than alteration of the genetic code itself.
	Literature: required and optional