

Course Unit Descriptor

Study program: MSc in Biology			
Course Unit Title: Medical genetics			
Course Unit Code: MB36			
Name of Lecturer(s): Nataša Kočiš Tubić, PhD			
Type and Level of Studies: Master's studies			
Course Status (compulsory/elective): elective			
Semester (winter/summer): winter			
Language of instruction: English			
Mode of course unit delivery (face-to-face/distance learning): face-to-face			
Number of ECTS Allocated: 7			
Requirements:			
Course Aimes: The aim of this course is to introduce students to the principles of diagnostics and the underlying mechanisms of hereditary disorders.			
Learning Outcomes: After successfully realized pre-exam and exam obligations student can understand the function of human genome and the importance of genetics for the origin of diseases in human populations, and procedure of screening and molecular diagnostics.			
Syllabus: <i>Theoretical instruction</i> Introduction to medical genetics. Chromosomal structure and number, normal and deviant karyotypes. Cytogenetics. Single-gene disorders. X-linked disorders. Polygenic and multifactorial inheritance. Biochemical genetics. Hemoglobinopathies. Pharmacogenetics. Immunogenetics. Cancer genetics. Screening for genetic diseases. Prenatal testing and techniques in prenatal diagnosis. Risk calculation. Gene therapy. Ethical issues in medical genetics. <i>Practical lecture</i> Solving problems of Medical Genetics. Pedigree analysis. Cytogenetics. Genotyping of monogenic disorders. Use of web based medical genetics databases and tools for genetic testing. Case-study problems. Journal Club.			
Literature Turnpenny P. Ellard S. Emery's elements of Medical Genetics. 13 Ed. Elsevier 2012			
Weekly teaching load	Lectures: 2	Teaching laboratory:	Other forms of teaching: 3
Teaching methods lectures, seminar, practical lecture, Journal Club, consultations			
Evaluation of knowledge (maximum score 100)			
Pre-exam obligation	points	Final exam	points
Seminar	Up to 20	Oral exam	Up to 70
Student engagement in practical lectures	Up to 10		