Faculty of Medicine / MEDICINE / CLINICAL GENETICS

Course:	CLINICAL GENETICS			
Course ID	**Course status**	**Semester**	**ECTS credits**	**Lessons** (Lessons+Exercises+Laboratory)
11141	Mandatory	10	4	2+2+0
Programs	MEDICINE			
Prerequisites	No prerequisites required			
Aims	The aim of the course is for students to master the principles of inheritance and the occurrence of hereditary disorders in humans, to become familiar with the types of genomic diseases, their clinical manifestations, the application of molecular genetics and recombinant DNA technologies in the diagnosis and treatment of genomic diseases in humans, and to master the basic principles of genetic counseling and applications of bioethical principles in clinical genetics.			
Learning outcomes	After completing the course the student will be able to: 1. Explain the organization of the human genome and the basic principles of clinical genetics and its application in modern diagnostics and therapy. 2. Recognize and describe types of hereditary diseases (chromosomal, monogenic, mitochondrial, multifactorial) and types of inheritance of monogenic diseases (autosomal-dominant, autosomal-recessive, X-dominant and X-recessive). 3. Describe and interpret the human karyotype and chromosomal aberrations using basic application of ISC Nomenclature. 4. Knows to list risk groups with a hereditary load in the population, to chose and define diagnostic methods, and to calculate the basic risks of recurrence. 5. Knows how to define and compare different types of genetic testing, argues the indications for individual genetic testing and uses available electronic databases of genetic data. 6. Knows the principles of prenatal diagnosis of hereditary diseases and knows how to state the criteria for differentiating high/low risk pregnancies, indications for invasive prenatal diagnosis and methods of invasive and non-invasive prenatal diagnosis of hereditary diseases. 7. Knows the basic principles and specifics of genetic counseling and providing genetic information and the basic bioethical principles of genetic data protection and indirect counseling.			
Lecturer / Teaching assistant	Full professor Olivera Miljanović, MD, PhD and associate assistant Jelena Jovanović, MD			
Methodology	Lectures, workshops, simulations, colloquia, seminars, exercises and consultations.			
Plan and program of work				
Preparing week	Preparation and registration of the semester			
I week lectures	Introduction to clinical genetics: History and impact of genetics on medicine science. Cellular and molecular basis of inheritance. The human genome.			
I week exercises	Introductory exercise – orientation on the principles of practical teaching			
II week lectures	Gene function. Chromosomes and cell division. ISC Nomenclature			
II week exercises	History of genomic load and diseases			
III week lectures	Developmental genetics.			
III week exercises	Recognizing the genomic risks			
IV week lectures	Models of inheritance: Mendelian, mitochondrial, non-Mendelian.			
IV week exercises	Evaluation of phenotypic characteristics in patient with genomic diseases			
V week lectures	Congenital anomalies. Malformation syndromes and dysmorphology.			
V week exercises	Dysmorphological examination: Recognition and registration of minor and major anomalies			
VI week lectures	Laboratory diagnostic methods in diagnostics of genomic disorders. Gene mapping and finding the causes of genomic diseases.			
VI week exercises	Introduction to principles of laboratory diagnostics.			
VII week lectures	Chromosomal diseases. Genetic counseling and genetic information.			
VII week exercises	Detection of minor malformations and assessment of dysmorphia score			
VIII week lectures	Gene mutations. The most significant monogenic diseases. Penetrance and gene expressivity, genetic heterogeneity.			
VIII week exercises	Karyotype interpretation - the HA electronic database			
IX week lectures	Hereditary metabolic diseases and neonatal screening			
IX week exercises	Principles of pedigree, Pedigree symbols and recognition the types of inheritance.			

X week lectures	Complex diseases: polygenic and multifactorial genetics. Population genetics, risk calculation for complex disorders.
X week exercises	Preparation of the pedigree for different types of hereditary diseases
XI week lectures	Intellectual deficiency, neurodevelopmental diseases and autism.
XI week exercises	Providing pre-testing genetic information and obtaining written consent.
XII week lectures	Prenatal diagnosis of hereditary diseases and congenital anomalies. Reproductive genetics and assisted reproductive technologies.
XII week exercises	Recognition of preconception and prenatal risks for genomics diseases.
XIII week lectures	Immunogenetics and Oncogenetics. Genetic testing in diagnostics i treatment of malignant diseases.
XIII week exercises	Interpretation of the results of prenatal diagnostics in the fetus.
XIV week lectures	Pharmacogenomics, personalized medicine and hereditary diseases treatment.
XIV week exercises	Interpretation of molecular genetic analyses. PCR, MLPA, aCGH, WES/CES.
XV week lectures	Genetic counseling, risk assessment and screening of people with high genomic risk.
XV week exercises	Writing and providing post-testing genetic information
Student workload	Classes and final exam: (5.33 hours) x 16 = 85.28 hours Necessary preparations before the beginning of the semester (administration, registration, certification): (5.33 hours) x 2 = 10.66 hours Total workload for the course: 4 x 30 = 120 hours Load structure: 85.28 hours (classes and final exam) + 10.66 hours (preparation) + 24.06 hours (supplementary work)

Per week	**Per semester**
4 credits x 40/30=5 hours and 20 minuts 2 sat(a) theoretical classes 0 sat(a) practical classes 2 excercises **1 hour(s) i 20 minuts** of independent work, including consultations	Classes and final exam: **5 hour(s) i 20 minuts x 16 =85 hour(s) i 20 minuts** Necessary preparation before the beginning of the semester (administration, registration, certification): **5 hour(s) i 20 minuts x 2 =10 hour(s) i 40 minuts** Total workload for the subject: **4 x 30=120 hour(s)** Additional work for exam preparation in the preparing exam period, including taking the remedial exam from 0 to 30 hours (remaining time from the first two items to the total load for the item) **24 hour(s) i 0 minuts** Workload structure: **85 hour(s) i 20 minuts (cources), 10 hour(s) i 40 minuts (preparation), 24 hour(s) i 0 minuts (additional work)**
Student obligations	Attending classes, actively participating in exercises and seminars, independent preparation of materials for seminars, solving set problems independently and in a group.
Consultations	After lectures, seminars and exercises, online and live in agreement with the professor and teaching assistant.
Literature	Literature: Turnpenny P, Ellard S. Emerjevi osnovi medicinske genetike. 13. izdanje (prevod sa engleskog jezika). Datastatus, Beograd 2009. ili Turnpenny P, Ellard S. Emery`s Elements of Medical Genetics. 15th ed. Elsevier, 2017. Additional literature (selected chapters): Nussbaum R.L, McInnes R.R, Willard H.E. Tompson & Tompson Genetics in medicine, Eight ed. Elsevier, 2016.
Examination methods	Knowledge assessment and grading: Class attendance: 10 points Seminar: 10 points Colloquium: 10 points Final exam: 70 points Grade: A B C D E F Points: : 90-100 80-89 70-79 60-69 50-59 < 50 Passed exam implies cumulatively accumulated at least 50 points
Special remarks	None
Comment	None

Grade:	F	E	D	C	B	A
Number of points	less than 50 points	greater than or equal to 50 points and less than 60 points	greater than or equal to 60 points and less than 70 points	greater than or equal to 70 points and less than 80 points	greater than or equal to 80 points and less than 90 points	greater than or equal to 90 points