MEDBIOT-06:	**MEDICAL GENETICS**
GENERAL INFORMATION	
Course lecturer	Marija Heffer, MD, PhD, full prof. with tenure
Associates	Jasenka Wagner, PhD, Assoc. Prof. Vedrana Ivić, PhD
Study programme	Interdisciplinary Graduate Study Programme in English: Biotechnology
Course status	Obligatory
Year of study, semester	2nd Year / 3rd Semester
Credit value (ECTS)	**4.5**
Teaching schedule (number of classes)	Lectures: 30; Seminars: 10; Exercises:15
Expected number of students for the course	25-30
DESCRIPTION OF THE COURSE	
Aims of the course	
The aim of the course is to gain knowledge in genomics, as a basic tool of precision medicine, which determines the risk of disease, response to therapy and suggests a lifestyle correction. Through learning about the structure of the genome, the consequences of genomic variation, multifactorial inheritance, epigenetic alterations, mitochondrial biology and pharmacogenomics, students will get an overview of the methods used to create large databases and bioinformatics tools for their diagnostic and scientific search.	
Requirements for course enrolment and starting competencies required for the course	
Completed undergraduate university study programme from the area of natural sciences (chemistry, biology) or biotechnical sciences, or biomedicine and healthcare. Completed and passed courses from 1st year of the study.	
Learning outcomes achieved at the level of the course programme	
MEDBIOT-2; MEDBIOT-3; MEDBIOT-5; BIOTECH-6; BIOTECH-7; BIOTECH-10	
Expected learning outcomes at the course level (5-10 outcomes)	
After completing the course the student will be able to: 1. Discern the cause-and-effect relationship between disease pathogenesis and changes at the gene and genome levels of an individual or population; 2. Link genome maintenance mechanisms to metabolic processes, physiological mechanisms, behavioural adaptations, and disease pathogenesis; 3. Identify genes, mutation sites and epigenetic changes that contribute to pathophysiological mechanisms of disease development, which may serve for diagnostic purposes or as targets of therapy; 4. Select appropriate genomic analyses required for research or diagnosis; 5. Critically evaluate the results of various genetic analyses and comment on valid evidence; 6. Find a suitable bioinformatics tool for analysing sequencing results, comparing two genomes or population analysis; 7. Comment the results of genomic scientific research.	
Course content	
Lectures. Factors in genetics, environment and lifestyle in the etiology of disease. Personalized medicines and therapeutic interventions. Integration of genomic and phenotypic data with digital tools to evaluate disease risk or monitor patient status. Epigenetic mechanisms, genomic reprogramming and inactivation of X chromosomes. Mutations in epigens. Genomic variability and congenital diseases. Mutations, insertions, deletions, and copy number variations. Inheritance patterns (recessive, dominant, sexually linked, penetrant). Statistics for geneticists. Connection	

analysis. Bioinformatics. Chromosomal anomalies. Unique inheritance. Multifactorial inheritance and complex diseases. HapMap and GWS. Population genetics. Comparative genomics. Pathogenesis of genomic diseases. Loci that accelerate or slow down aging. Progeria. Genetic counselling and bioethics.

Seminars. Nature and frequency of hereditary diseases in various parts of the world. Genomic imprinting. Cancer epigenetics. Environmental impact on the occurrence of epigenetic markers. Genotype and phenotype correlation. Searching for genomic bases. Mitochondrial inheritance. Gene therapy of mitochondrial diseases. Inheritance calculation and risk assessment within families and populations.

Exercises. Preparation and submission of DNA samples for various genomic analyses and assays. Isolation of DNA samples from biological traces, very old or contaminated samples. Family trees from genomic analyses. DNA sequencing techniques. The methods for SNP detection. Methods for the analysis of epigenetic markers. Techniques for determining chromosomal anomalies. Methods for mitochondrial DNA analysis.

Types of classes

Lectures; Seminars; Laboratory exercises

Student obligations

Attendance at all forms of classes is mandatory and the students are obligated to attend all knowledge tests. The students may be absent from 30% (full-time students) and 50% (part-time students) of each of the forms of classes, provided that the absence is justified. An exercise or a seminar which has not been completed must be made up through a midterm exam.

Monitoring the students' work *(Connecting learning outcomes, teaching methods, and grading)*

Teaching activity	ECTS	Learning outcome	Student activities	Evaluation methods	Grade points	
					Min.	Max.
Attendance (lectures, seminars, Exercises)	1	1-7	Presence in class, Seminar paper Practical work	Record	1 10 12	5 20 25
Knowledge assessment (partial tests)	1	1-7	Learning for a partial test	Written exam	10	20
Final exam	2.5	1-7	Learning for the final exam	Written exam	18	30
Total	**4.5**				**50**	**100**

Table 2. Evaluation of the written part of the final exam

Percentage of correct answers (%)	Grade
>95.00	30
90.00-94.99	29
85.00-89.99	28
80.00-84.99	26
75.00-79.99	24
70.00-74.99	22
65.00-69.99	20
60.00-64.99	18

Forming the final grade:

The points granted for the final exam are added to the grade points awarded during class attendance. The grading process is conducted by absolute distribution, i.e. based on total

achievements, and compared to the numerical system in the following manner:
A – Excellent (5): 90-100 grade points; B – Very Good (4): 80-89.99 grade points; C – Good (3): 65-79.99 grade points; D – sufficient (2): 50-64.99 grade points

Mandatory course literature (available at the library and in other media)

Title	Number of copies in the library	Availability in other media
Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics. Foundation, Academic Press, 7th Ed., 2019.	10	

Additional course literature

Turnpenny PD, Sian E: Emery's Elements and Medical Genetics. Student Consult. Elsevier, 2017.

Methods for monitoring quality that ensure the acquisition of outcomes and competencies

Anonymous, quantitative, standardised student survey on the course and the teacher's work implemented by the Quality improvement office of the Faculty of Medicine Osijek and/or the Faculty of Food Technology Osijek.

Note

E-learning is not within the standard of the class, but is used in teaching and contains links to various pages, videos and audio materials available on the web pages.