BIOTECH-03:	**GENETICS AND GENOMICS**
GENERAL INFORMATION	
Course Coordinator(s)	Stana Tokić, PhD, assist. prof.
Associate(s)	Mario Štefanić, MD, PhD, assoc. prof. Jasminka Wagner, PhD, assoc. prof. Teuta Opačak-Bernardi, PhD, assist. prof. Barbara Viljetić, PhD, assist. prof.
Study Programme	Interdisciplinary Graduate Study Programme in English: Biotechnology
Course Status	Obligatory
Year of Study, Semester	1st Year / 1st Semester
Credits (ECTS)	**7**
Teaching Method (number of classes)	Lectures: 45; Seminars: 15; Exercises: 15
Expected Number of Students in the Course	25-30
COURSE DESCRIPTION	
Course Aims	
The objective of the course is to broaden and improve the basic knowledge of classical genetics and genomics by applying various methods and bioinformatics tools that students can use in biological research and molecular biotechnology.	
Prerequisites for Enrolment and the Entry 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.	
Learning Outcomes at the Programme Level Contributed by the Course	
MEDBIOT-1; INDBIOT-2; BIOTECH-7; MEDBIOT-3	
Learning Outcomes at the Course Level	
After completing the course the student will be able to: 1. Present the principles of inheritance 2. Apply practical knowledge for qualitative and quantitative genome analysis 3. Select appropriate statistical methods and bioinformatics tools for genome and transcriptome research 4. Assess the role of epigenetic factors in the development of complex diseases 5. Apply classical and molecular cytogenetic techniques and approaches in the diagnostics of hereditary diseases 6. Follow the scientific and professional literature and improve the acquired knowledge and skills	
Course Content	
Lectures. Model organisms - history, types and applications in genome research. Gene mapping and cloning techniques - classic gene crossing and complementation, restriction endonucleases, plasmids (pBR322, pUC19), bacteriophages (λ, M13) and cosmids, replication and expression vectors. Strategies for gene marking, genetic material transfer and genetic library screening. Numerical and structural chromosomal anomalies. Principles of inheritance: monogenic and polygenic, Mendelian laws of inheritance and allele types. Sex-linked and mitochondrial inheritance. Complications of the basic Mendelian mode of inheritance: incomplete penetration and gene expression, anticipation, pleiotropy, gene conversion, epistasis, genomic imprinting, mosaicism and chimerism. Population genetics: allelic and gene frequencies. Hardy-Weinberg equilibrium. Linkage disequilibrium (LD). Gene polymorphisms, haplotypes, LD blocks and taqSNPs. HapMap project. Quantitative trait loci (QTL). Linkage analyses in search of genes causing hereditary diseases - LOD and λ linkage measures. Genome wide association studies (GWAS). Factors shaping the genetic characteristics of a population - genetic drift, natural selection, inbreeding. Tools for human	

genome variability assessment and gene mapping - PCR (multiplex, nested, reverse-transcription, real-time, touchdown, asymmetric), RFLP, SNP and STR analysis, FISH chromosome mapping, genomic library search (BAC and YAC) and positional cloning, contig and restriction maps, candidate gene analysis, UCSC Human Genome Viewer. Evolution of DNA sequence analysis – Maxam and Gilbert method, Sanger method, next generation sequencing (NGS) - overview of experimental protocols and NGS platforms (Illumina, Roche, SOLiD, Ion Torrent, Nanopore, PacBio), and data analysis in comparison to reference genomic databases. Methods for gene expression (qPCR, microarray and mRNA / microRNA sequencing) and gene function analysis (knockout / knockdown transgenic animal models). The role of epigenetic factors in regulation of gene expression and disease occurrence. Clinical genetics and diagnostics of genetic diseases - cytogenetic testing, risk allele genotyping, predictive and prenatal testing, DNA profiling, genetic counselling and a personalized approach to therapy.

Seminars. Model organisms in biotechnology research. Principles and application of statistical methods in genetics. Whole-exome NGS analysis. Application of chromosomal microarray in diagnostics. Application of pharmacogenetics in personalized therapy.

Exercises. DNA isolation by affinity method, fragmentation by restriction enzymes and restriction fragment length polymorphism (RFLP) analysis using agarose gel electrophoresis. Qualitative and quantitative DNA analysis by agarose gel electrophoresis, Qubit fluorometer and qPCR analysis. STR analysis by capillary electrophoresis. RNA isolation, cDNA synthesis and gene expression analysis using TaqMan technology and RT-qPCR method. Chromosome karyotyping of peripheral mononuclear leukocytes by GTG banding. Detection of microdeletion syndromes by FISH method.

Teaching Methods

Lectures; seminars; laboratory exercises

Students' 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 Activity of the Students *(Connecting Learning Outcomes, Teaching Methods, and Grading)*

Class-related activity	ECTS	Learning outcome	Student activity	Evaluation method	Grade points	
					Min.	Max.
Attending and active participation (lectures)	0.5	1-6	Attendance at classes	Keeping records	2	10
Seminar work	1	1-6	Seminar work	Presentation of seminar work	10	20
Laboratory exercises	1	5,6	Work in laboratory	Written report	8	20
Final exam	3.5	1-6	Studying for the final exam	Written exam	30	50
Total	**6**				**50**	**100**

Evaluation of the written part of the final exam

Percentage of correct answers (%)	Grade
>95.00	50
90.00-94.99	47
85.00-89.99	45
80.00-84.99	40
75.00-79.99	38
70.00-74.99	35
65.00-69.99	33
60.00-64.99	30

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 Literature (available in the library and via other media)

Title	Number of copies in the library	Availability via other media
Strachan T, Read A: Human Molecular Genetics. 4th Edition. Wiley, 2003.	-	
Dale JW, von Schantz M: From Genes to genomes. John Wiley & Sons, LTD. University of Surrey, UK, 2002.	-	
Scientific and professional papers related to particular chapters (available online)		Yes

Additional Literature

M.R. Green, J. Sambrook, Molecular Cloning: A Laboratory Manual, 4th Ed., Cold Spring Harbor Laboratory Press, New York, 2012.

Quality Assurance Procedures Designed to 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 included in the class quota, but it is used in teaching and it contains links to various sites and video and audio materials available on websites.