MOLECULAR GENETICS - FROM SCIENTIFIC HYPOTHESIS TO MEDICAL PRACTICE	
GENERAL INFORMATION	
Course coordinator	Professor Marija Heffer, MD, PhD
Assistant/Associate	Asst. Prof. Vedrana Ivić, PhD Marta Balog, PhD
Study Programme	Integrated undergraduate and graduate university study of Medicine
Status of the course	Elective
Year of study, semester	1th year, 2nd semester
ECTS	**2**
Workload (hours)	Lectures (5); Seminars (20)
Expected number of students	30
COURSE DESCRIPTION	
Course objectives	
Link the structure and function of the human genome and mitochondrial DNA to the selection mechanisms that shaped them (environment, diet, infectious organisms) and the mechanisms that affect them during development and adulthood (genotoxic and epigenetic influences) in order to develop analytical thinking about molecular diagnostics and therapies.	
Enrolment requirements and entry competencies	
Completed the course Medical Biology.	
Learning outcomes at the Programme level	
Learning outcomes (5-10)	
After completing the course, the student will be able to: 1. Analyze the structure of the human genome and bring it into the context of historical migrations, selective environmental pressures, changing dietary styles and exposures to infectious organisms. 2. To connect the structure and function of mitochondrial DNA with the efficiency of energy production, the spread of mitochondrial haplotypes in climatic zones, the mechanisms that contribute to aging and the development of chronic diseases. 3. Integrate genetic and epigenetic approaches in explaining the pathogenesis of human diseases, find reliable sources of genetic data related to individual diseases in large databases and analyze the possibilities of therapeutic approaches. 4. Explain the possibilities of diagnosis and therapeutic genetic interventions in reproduction, metabolic and tumor diseases. 5. Describe aging through mechanisms that act on the genome, explain and critically review existing senescence therapeutic interventions and methods of measuring biological age.	
Course content	
Lectures Genetic record of the evolution of the human species. Evolution of sex chromosomes - will the Y chromosome disappear? Regulation of gene expression - from DNA to proteins. Integration of epigenetic and genetic approaches of pathogenesis of human diseases. Genetics of aging. **Seminars** Selective pressure of infectious organisms on the evolution of the human genome. Gene polymorphism. Organization, mode of inheritance and determination of hereditary lineage	

according to mitochondrial DNA. Association of aging and chronic diseases with mitochondrial DNA. Functional and comparative genomics. Tumor genetics. Pharmacogenomics. Therapeutic genetic interventions. Genetically modified organisms in the service of modern medicine. Reproductive genetics.

Mode of teaching

Lectures; Seminars

Student obligations

Attendance at all forms of classes is mandatory, and the student must access all knowledge tests. A student may justifiably miss 30% of each form of instruction. Unfinished exercise must be colloquial.

Monitoring student work *(alignment of learning outcomes, teaching methods and grading)*

Teaching activity	ECTS	Learning outcome	Student activity	Assessment methods	Grade points	
					Min.	Max.
Lectures	0,25	1-5	Class attendance	Record	6	12
Seminars	0.25	1-5	Participation in the discussion	Tracking activity	6	12
Essay writing	0.75	1-5	Writing an essay on a selected topic	Evaluation of a written essay	19	38
Final exam	0.75	1-5	Learning for the final exam	Written exam	19	38
Total	**2**				**50**	**100**

Calculation of final grade:
Students who achieved 19 or more points in the final exam, the points obtained in the final exam are added to the grade points obtained during the class, and this sum constitutes the final grade. Since the study program schedule descriptive assessment of elective courses, the course coordinator awards the grade "passed" to a student who achieves 50 or more grade points in the course.

Required reading (available in the library and through other media)

Title	Number of copies in the library	Availability through other media
1. Geoffrey M. Cooper i Robert E. Hausman: Stanica-Molekularni pristup, 5. izdanje, Medicinska naklada, Zagreb, 2010.	14	

Additional reading

Selected papers:
1. The ENCODE Project Consortium., Moore, J.E., Purcaro, M.J. *et al.* Expanded encyclopaedias of DNA elements in the human and mouse genomes. *Nature* **583,** 699–710 (2020). https://doi.org/10.1038/s41586-020-2493-4
2. Spyrou, M.A., Bos, K.I., Herbig, A. *et al.* Ancient pathogen genomics as an emerging tool for infectious disease research. *Nat Rev Genet* **20,** 323–340 (2019). https://doi.org/10.1038/s41576-019-0119-1

3. Yongjun L., Xiaohong Y., Yugi G. Mitochondrial DNA response to high altitude: A new perspective on high-altitude adaptation, Mitochondrial DNA 24(4), 313-319 (2013).
4. Turajlic, S., Sottoriva, A., Graham, T. *et al.* Resolving genetic heterogeneity in cancer. *Nat Rev Genet* **20,** 404–416 (2019). https://doi.org/10.1038/s41576-019-0114-6
5. Daly AK. Pharmacogenetics: a general review on progress to date. Br Med Bull. 2017 Dec 1;124(1):65-79. doi: 10.1093/bmb/ldx035. PMID: 29040422.

Course evaluation procedures

Anonymous, quantitative, standardized student survey on the subject and work of teachers conducted by the Office for Quality of the Medical Faculty Osijek.

Note /Other

E-learning is not included in the norm of subject hours, but it is used in teaching and contains links to various pages, video and audio materials available on the website.