COURSE UNIT (MODULE) DESCRIPTION

Course unit title	Code
Human genetics	

Lecturer(s)	Department(s)
Coordinating: Birutė Tumienė, assoc.prof., MD, PhD Others: lecturers from the Department of Human and Medical Genetics, Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University	Department of Human and Medical Genetics, Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University, Santariškių str. 2, Vilnius

Cycle	Type of the course unit
Integrated studies (I and II cycles)	Compulsory

Mode of delivery	Period of delivery	Language of instruction
Lectures, practicals and self-study	Semester III (autumn)	English

Prerequisites and corequisites	
Prerequisites: General knowledge of natural sciences gained with secondary education	**Corequisites (if any):** None

Number of ECTS credits allocated to the course unit	Total student's workload	Contact hours	Self-study hours
2	54	26	28

Purpose of the course unit (module): programme competences to be developed		
To provide knowledge about the general principles of human genetics and the impact of genetic factors on human health and disease. To explain the principles of genetic testing, their differences, specific applications, interpretation of test results, and their role in the diagnosis of genetic diseases.		
General learning outcomes of the course unit	**Teaching and learning methods**	**Assessment methods**
The student will conduct oneself honestly and adhere to ethical commitments. Will be able to think critically and self-critically, will be creative, proactive will strive to achieve ones objectives, will be able to communicate and work in a team.	Problem-oriented lecturing and demonstrations, active learning methods (group discussions), analytical methods (information search and analysis)	Continuous evaluation during the semester; exam- test (open and closed type tasks/questions)
Students will be able to evaluate the limits of their competences and if needed seek assistance. Will be able to solve problems and make decisions. Collaborate with specialists and scientists from other areas and disciplines.	Active learning methods (group discussions)	Continuous evaluation during practicals; exam
Academic learning outcomes of the course unit		
The student will demonstrate knowledge about human genome structure, inheritance and realization of genetic information, genetic mutability and its outcomes.	Problem-oriented lecturing and demonstrations, active learning methods (group discussions), analytical methods (information search and analysis)	Continuous evaluation during the semester;
The student will understand the contribution of the genetic factors to human ontogenesis, health and		

disease and will be able to draw a pedigree and to define the mode of inheritance.		exam- test (open and closed type tasks/questions)
The student will be able to search for relevant information about human inherited traits, diseases and related genetic factors independently.		
The student will be able to explain the main principles of the basic genetic analysis methods, their clinical applications, analysis and results in relation with clinical symptoms.		

Topics	Contact work hours							Time and tasks of self-study	
	Lectures	Consultations	Seminars	Practice	Laboratory work	Practical training	**Total contact hours**	**Self-study**	**Tasks**
1. Structure of the DNA and human genome organization. Structure and variation of human genes. Molecular and morphological structure of human chromosomes and karyotyping. The cell cycle, similarities and differences between mitosis and meiosis, genetic recombination, and its biological significance.	2						2	2,5	Independent analysis of indicated literature, seminars and preparation for the exam
2. Mutational variability: mutations, polymorphisms, genetic variants. Types of mutations, their formation mechanisms, and clinical consequences. Mutagenesis – causes of mutations, groups of mutagens. DNA damage repair mechanisms.	2						2	2,5	
3. Genomic mutations: types and consequences. Changes in chromosome numbers and common syndromes. Features of human gamete formation. Genetic mosaicism and chimerism. Balanced and unbalanced chromosomal structural rearrangements.	2						2	2,5	
4. Gene expression mechanisms and regulation. Genomic imprinting and imprinting disorders.	2						2	2,5	
5. Monogenic inheritance: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, Y-linked (holandric). Monogenic diseases.	2						2	2,5	
6. Factors in disturbance of basic Mendelian patterns: penetrance, variable expression, pleiotropy. Non-Mendelian patterns of inheritance, mitochondrial diseases. Somatic mutations, cancer genetics. Multifactorial inheritance, contribution of genetic factors to common diseases. Oligogenic and polygenic inheritance of human traits, quantitative genetics.	2						2	2,5	
7. Developmental genetics: stem cells, cell differentiation, morphogenesis. Developmental anomalies. Teratogenesis.	2						2	2,5	
8. Concept of inheritance, genetic factors in human health and diseases. Genetics and genomics in contemporary medicine. Collection of family history and pedigree drawing. Genetic counselling: objectives, principles, process. Assessment of	2						2	2,5	

genetic risks. Bioethical issues in genetic counselling.									
9. Population genetics: genetic variation of populations, application of Hardy-Weinberg equation to allele frequency calculations in populations. Evolutionary genetics.	2						2	2,5	
10. Molecular genetics: introduction to molecular genetics methods. Object and subject of genetic analyses, polymerase chain reaction. Application of molecular genetic methods for the identification of DNA and chromosomal number rearrangements.				4			4	3	Preparation for the practicals according to materials given during lectures
11. Cytogenetics: nomenclature of chromosomal mutations according to ISCN. Application of routine staining (G-banding) for the analysis of chromosomes – formation of karyogram and identification of mutations.				4			4	2,5	
Consultation									
Total	**18**			**8**			**26**	**28**	

Assessment strategy	**Weight (%)**	**Assessment period**	**Assessment criteria**
Human genetics practicals	Passed	During the semester	All tasks in practical classes should be evaluated (completed/not completed) by the end of the semester. Students will be allowed into the exam only in case all tasks are completed.
Human genetics task: Pedigree	10%	During the semester	Pedigree, completed according to requirements and presented to lecturer in time. Students will be allowed into the exam only in case the pedigree task is completed and passed. Evaluation: 10: Excellent knowledge and abilities. Level of evaluation and synthesis of the facts and outcomes. 9: Very good knowledge and abilities, minor mistakes might occur. 8: Good knowledge and abilities, with minor mistakes. 7: Average knowledge and abilities, various mistakes occur. 6: Below average knowledge and abilities with major mistakes. 5: Satisfactory (minimum) knowledge and abilities. Many mistakes. 0-4: Knowledge and abilities are below satisfactory level. Not passed.
Human genetics colloquium	20%	During the semester	Each colloquium consists of 20 short tasks/questions from the course of human genetics lectures (open and closed type). The final mark is attributed as follows: 10: Excellent knowledge and abilities. Level of evaluation and synthesis of the facts and outcomes. 100-91% correct answers. 9: Very good knowledge and abilities, minor mistakes might occur. 90-82% correct answers. 8: Good knowledge and abilities, with minor mistakes. 81-73% correct answers. 7: Average knowledge and abilities, various mistakes occur. 72-64% correct answers. 6: Below average knowledge and abilities with major mistakes. 63-55% correct answers. 5: Satisfactory (minimum) knowledge and abilities. Many mistakes. 54-46% correct answers. 0-4: Knowledge and abilities are below satisfactory level. <45% correct answers. Failed. Students will be allowed into the exam only in case the colloquium is passed.
Examination	70%	Session period	Students will be allowed to take the exam only in case the human genetics pedigree task, colloquium and both practicals are passed.

			Exam consists of 20 short tasks/questions from the whole course of Human genetics. The final mark is attributed as follows: 10: Excellent knowledge and abilities. Level of evaluation and synthesis of the facts and outcomes. 100-91% correct answers. 9: Very good knowledge and abilities, minor mistakes might occur. 90-82% correct answers. 8: Good knowledge and abilities, with minor mistakes. 81-73% correct answers. 7: Average knowledge and abilities, various mistakes occur. 72-64% correct answers. 6: Below average knowledge and abilities with major mistakes. 63-55% correct answers. 5: Satisfactory (minimum) knowledge and abilities. Many mistakes. 54-46% correct answers. 0-4: Knowledge and abilities are below satisfactory level. <45% correct answers. Failed.
The final cumulative grade		During the semester and session	The final cumulative grade will be obtained according to the formula: human genetics colloquium, 20%; K (0,2 X K) + human genetics pedigree task 10%; G (0,1 X G) + examination, 70%; E (0,7 X E): 0,2 X K + 0,1 X G + 0,7 X E.

Author	**Year of publication**	**Title**	**No. of periodical or vol. of publication**	**Publication place and publisher or internet link**
Required reading				
G. Bradley Schaefer, James N. Thompson, Jr.	2014	Medical Genetics: An Integrated Approach		
Dennis Kasper, Anthony Fauci, Stephen Hauser, Dan Longo, J. Larry Jameson, Joseph Loscalzo	2015	Harrison'sTM Principles of Internal Medicine	19th ed.	
William W. Hay, Jr., Myron J. Levin, Robin R. Deterding, Mark J. Abzug	2018	Current Diagnosis & Treatment: Pediatrics	24th ed.	
On-line database: Online Mendelian Inheritance in Man (OMIM)				https://www.ncbi.nlm.nih.gov/omim
On-line database: Ensembl				https://www.ensembl.org/index.html
On-line database: BLAST				https://blast.ncbi.nlm.nih.gov/Blast.cgi
Recommended reading				
Open-source on-line resources	http://www.eurogems.org/ http://opengenetics.net/index.html https://www.broadinstitute.org/scientific-community/science/programs/medical-and-population-genetics/primers/primer-medical-and-pop http://www.ygyh.org/			

	https://www2.le.ac.uk/projects/vgec