



COURSE UNIT (MODULE) DESCRIPTION

Course unit title	Code
Human biology and genetics	BZBI1112

Lecturer(s)	Department(s)
Coordinating: Rimantas Jankauskas, prof., PhD, Others: Algirdas Utkus, prof., MD, PhD Violeta Bartuškienė, Lect., PhD Birutė Tumienė, assist., MD, PhD Vytautas Šliužas, assist., PhD Karolis Baronas, lect.	Department of Anatomy, Histology and Anthropology, Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University, M.K. Čiurlionio str. 21, Vilnius Department of Human and Medical Genetics, Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University, Santariškių str. 2, Vilnius

Cycle	Level of the course unit	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 I (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
5	134	67	67

Purpose of the course unit (module): programme competences to be developed		
The objective of the course is to consolidate the general knowledge of biology and genetics by studying the main object of the study program - the human being as a special biological and social phenomenon.		
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		
With the use of the knowledge of general biology the student will be able to critically assess the information about a human being and the natural world around him and will be able to suggest adequate solutions to problems.	Problem-oriented lecturing and demonstrations, active learning methods (group discussions, collaboration based teamwork), analytical methods (information search and analysis)	Continuous evaluation during the semester; exam- test (open and closed type tasks/questions)
The student will be able to independently gain knowledge about the human being and its biotic and social environment. Will be able to find, evaluate and systematize information about a chosen organism.	Problem-oriented lecturing and demonstrations, active learning methods (group discussions),	Continuous evaluation during the semester;

	analytical methods (information search and analysis)	exam- test (open and closed type tasks/questions)
The student will be able to demonstrate the knowledge about the peculiarities of human biology (phylogenesis, ontogenesis, human biological diversity and its causes). Will understand the effect of biological evolution factors to human health and disease.	Problem-oriented lecturing and demonstrations, active learning methods (group discussions), analytical methods (information search and analysis)	Continuous evaluation during the semester; Preparation and presentation of a group project based on literature analysis; exam- test (open and closed type tasks/questions)
The student will demonstrate knowledge about human genome structure, inheritance and realization of genetic information, genetic changeability and its outcomes.	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)
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.		
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. Human biology and anthropology. Principals of scientific research in natural sciences. Specific methods in anthropology. Genetic balance within human populations and the factors that affect it. The characteristics of primate groups, their morphological, physiological and behavioral adaptations. The key stages of anthropogenesis: primates of the third period; Pliocene hominids; Biological characteristics of <i>Homo erectus</i> and Acheul culture; forms of archaic humans, Neanderthals and Moustier culture. The hypothesis of the origins of modern human. The ecological and demographic characteristics as well as health and disease during the hunter-gatherer, early agriculture, early urbanization and industrial periods.	8						8	8	Independent analysis of indicated literature and preparation for the exam
2. Developmental biology (ontogenesis). The peculiarities of the human postnatal ontogenesis (physical, intellectual and social development). The peculiarities of physical and psychomotor development in neonatal period and childhood: the principals of sexual pubescence; morphological,	6						6	6	Independent analysis of indicated literature and preparation for the exam

physiological and social aspects of aging. Factors affecting ontogenesis. Human ageing and lifespan. Evolutionary factors in human ontogenesis.									
3. Anthropometric measurements and proportions of the modern human beings. Body composition and constitution, morphological, physiological and psychological aspects of constitution. The principles of human adaptation and acclimatization. Ecological gradients. The global diversity of human quantitative and discrete traits. The interpretations of the biological diversity of the modern human being (typological and evolutionary views). The effect of evolutionary factors on human diversity.	4						4	4	Independent analysis of indicated literature and preparation for the exam
4. Principles of evolutionary medicine. The understanding of human disease from the evolutionary perspective (reproduction, nutrition and metabolism, defence mechanisms and behaviour).	6			8			14	15	Independent analysis of indicated literature, preparation and presentation of a group project based on literature analysis; and preparation for the exam
1. Concept of inheritance, genetic factors in human health and diseases. Genetics and genomics in contemporary medicine. Collection of family history and pedigree drawing.	2						2	6	Independent analysis of indicated literature, seminars and preparation for the exam
2. Structure of the DNA and human genome organization. Structure and variation of human genes. Molecular and morphological structure of human chromosomes and karyotyping.	2						2	3	
3. Cell cycle, its control and regulation. Cell proliferation – mitosis and meiosis, stages and differences. Genetic recombination process and its biological significance. Formation of the male and female gametes.	2						2	3	
4. Mutational variability: mutations, polymorphisms, genetic variants. Variety and outcomes of the mutations. Mutagenesis and DNA repair.	2						2	2	
5. Genomic mutations: types and consequences. Numerical chromosomal abnormalities and related syndromes. Genetic mosaics and chimera. Balanced and unbalanced chromosomal structural rearrangements.	2						2	2	
6. Monogenic inheritance: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, Y-linked (holandric). Monogenic diseases.	2						2	2	
7. Factors in disturbance of basic Mendelian patterns: penetrance, variable expression, pleiotropy. Non-Mendelian patterns of inheritance, mitochondrial diseases. Somatic mutations, cancer genetics.	2						2	2	
8. Multifactorial inheritance, contribution of genetic factors to common diseases. Oligogenic and polygenic inheritance of human traits, quantitative genetics.	2						2	2	
9. Gene expression mechanisms and regulation. Genomic imprinting and imprinting disorders.	2						2	2	
10. Developmental genetics: stem cells, cell differentiation, morphogenesis. Developmental anomalies. Teratogenesis.	2						2	2	

11. Genetic counselling: objectives, principles, process. Assessment of genetic risks. Bioethical issues in genetic counselling.	2					2	2	
12. Population genetics: genetic variation of populations, application of Hardy-Weinberg equation to allele frequency calculations in populations. Evolutionary genetics.	2					2	2	
13. 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.			4,5			4,5	2	Preparation for the practicals according to materials given during lectures
14. 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.			4,5			4,5	2	
Consultation		2						
Total Human biology	24	1	8			33	33	
Total Human genetics	24	1	9			34	34	
Total	48	2	17			67	67	

Assessment strategy	Weight (%)	Assessment period	Assessment criteria
Human biology practicals: presentation of the group project	15%	During the semester	<p>Presentations delivered during the practicals: an in advance prepared presentation of the group project, based on the analysis of the scientific literature. The quality of the content, the presentation of scientific ideas, the validity of the conclusions, the quality of the scientific sources used, the structure and clarity of the report, the questions and answers of the audience are evaluated by the 4-point system:</p> <p>4: Great presentation of the group project: clear scientific ideas, smooth structure of the presentation, detailed analysis, systematic information, number and quality of scientific literature sufficient for credible analysis and conclusions, effective management of the audience.</p> <p>3: Good presentation of the group project: clear scientific ideas, clear presentation structure, conclusions are well presented, but information lacks generalization and structuring.</p> <p>2: satisfactory presentation of the group project: mostly clear scientific ideas, the presentation includes all required components, but the information lacks generalization and structuring, conclusions are not completely substantiated, minor errors are encountered, not all aspects of discussion are clear to the audience.</p> <p>1: weak preparation and presentation of the group project: scientific ideas revealed incompletely, vague structure of the presentation, incomplete analysis, conclusions lack validity and generalizations, insufficient scientific literature, sources of dubious scientific value dominate, insufficient interest of the audience.</p>
Human biology practicals	Passed	During the semester	Human biology practicals: participation in practicals and constructive engagement in the discussions, presentation of a group project. Absent in not more than one human biology practical.
Human genetics practicals	Passed	End of 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 practicals: Pedigree		End of the semester	Pedigree, completed according to requirements and presented to lecturer in time. Students will be allowed into the exam only in case pedigree is completed and presented in time.
Human genetics practicals: Colloquium (2)	15%	During semester	Each colloquium consists of 20 short tasks/questions (open and closed type), each response equals to 0,5 point. The final mark is attributed as follows: <u>10</u> : Excellent knowledge and abilities. Level of evaluation and synthesis of the facts and outcomes. 18-20 correct answers. <u>9</u> : Very good knowledge and abilities, minor mistakes might occur. Level of facts evaluation. 15-17 correct answers. <u>8</u> : Good knowledge and abilities, with minor mistakes. Level of facts synthesis. 12-14 correct answers. <u>7</u> : Average knowledge and abilities, various mistakes occur. Level of facts analysis. 10-11 correct answers. <u>6</u> : Below average knowledge and abilities with major mistakes. Level of knowledge application. 8-9 correct answers. <u>5</u> : Satisfactory (minimum) knowledge and abilities. Many mistakes. Level of facts understanding. 6-7 correct answers. <u>0-4</u> : Knowledge and abilities are below satisfactory level. 0-5 correct answers. Students will be allowed into the exam only in case both colloquiums are passed.
Examination	70%	Session period	Students will be allowed to take the exam only in case the human biology practicals are passed, human genetics pedigree is completed and presented in time, both colloquiums and practicals are passed. Exam consists of 40 short tasks/questions (open and closed type). Each response equals to 0,25 point. The final mark is attributed as follows: <u>10</u> : Excellent knowledge and abilities. Level of evaluation and synthesis of the facts and outcomes. 36-40 correct answers. <u>9</u> : Very good knowledge and abilities, minor mistakes might occur. Level of facts evaluation. 32-35 correct answers. <u>8</u> : Good knowledge and abilities, with minor mistakes. Level of facts synthesis. 28-31 correct answers. <u>7</u> : Average knowledge and abilities, various mistakes occur. Level of facts analysis. 24-27 correct answers. <u>6</u> : Below average knowledge and abilities with major mistakes. Level of knowledge application. 20-23 correct answers. <u>5</u> : Satisfactory (minimum) knowledge and abilities. Many mistakes. Level of facts understanding. 16-19 correct answers. <u>0-4</u> : Knowledge and abilities are below satisfactory level (unsatisfactory). 0-15 correct answers.

Author	Year of publication	Title	No. of periodical or vol. of publication	Publication place and publisher or internet link
Required reading				
Pollard TM.	2008	Western Diseases: an Evolutionary Perspective.		Cambridge University Press, https://www.cambridge.org/core/books/western-diseases/2DB8074F99CE69BE8964EA85129B2FEB
Stinson S., Bogin B., O'Rourke D.H. (ed.)	2012	Human biology: an evolutionary and biocultural perspective.	2 nd ed.	https://ebookcentral.proquest.com/lib/viluniv-

				ebooks/reader.action?docID=693178
Gluckman P., Beedle A., Hanson M.	2009 - 2012	Principles of evolutionary medicine.		Oxford University Press, https://ebookcentral.proquest.com/lib/viluniv-ebooks/detail.action?docID=472090
Gluckman P., Hanson M.	2006	Mismatch: why our world no longer fits our bodies.		https://ebookcentral.proquest.com/lib/viluniv-ebooks/reader.action?docID=430466
Karp G.	2013	Cell and molecular biology: concepts and experiments.	7 th ed.	John Wiley and Sons, USA
Tumienė B., Šliužas V., Baronas K.	2018	Conspect of human and medical genetics		https://vma.esec.vu.lt/
G. Bradley Schaefer, James N. Thompson, Jr.	2014	Medical Genetics: An Integrated Approach		https://accessmedicine.mhmedical.com/book.aspx?bookid=2247
Dennis Kasper, Anthony Fauci, Stephen Hauser, Dan Longo, J. Larry Jameson, Joseph Loscalzo	2015	Harrison's TM Principles of Internal Medicine	19 th ed.	https://accessmedicine.mhmedical.com/book.aspx?bookid=1130
William W. Hay, Jr., Myron J. Levin, Robin R. Deterding, Mark J. Abzug	2018	Current Diagnosis & Treatment: Pediatrics	24 th ed.	https://accessmedicine.mhmedical.com/book.aspx?bookid=2390
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				
Muehlenbein M.P.	2010	Human evolutionary biology.		Cambridge University Press
Stanford C., Allen J.S., Anton S.C.	2013	Biological anthropology: the natural history of humankind.		Pearson Education, Inc.
Jurmain R., Kilgore L., Trevanthen W,	2009	Essentials of Physical Anthropology.	7 th ed.	Wadsworth
Chiras D.D.	2015	Human biology.	8 th ed.	Jones and Bartlett Learning, USA
Cartwright J.	2000	Evolution and human behaviour.		Bradford Books
Strachan T, Read A.P.	2010	Human Molecular Genetics.	4 th ed.	Garland Publishing, USA

I.D. Young I.D.	2006	Introduction to risk calculation in genetic counselling.	3rd ed.	Oxford University Press
Strachan T., Read A.P. (eds.)	2010	Human Molecular Genetics.	4th ed.	Garland Publishing, JAV
Turnpenny P., Ellard P. (eds.)	2012	Emery's elements of medical genetics	14th ed.	Churchill Livingstone Elsevier, Philadelphia, JAV.
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			