Course unit (module) title	Code
Genomics	

Lecturer(s)	Department(s) where the course unit (module) is delivered
Coordinator: prof. dr (HP) Algirdas Utkus **Other(s): prof. dr. Audronė Jakaitienė, dr. Tautvydas Rančelis, dr. Beata Aleksiūnienė, dr. Ingrida Domarkienė, dr. Violeta Mikštienė, Evelina Siavrienė, Karolis Šablauskas**	Department of Human and Medical Genetics Vilnius University Faculty of Medicine, Centre for Medical Genetics Santaros Clinics Santariškių str. 2, Vilnius

Study cycle	Type of the course unit (module)
Second cycle	Compulsory

Mode of delivery	Period when the course unit (module) is delivered	Language(s) of instruction
Distance learning, face-to-face, self-study Lectures, seminars and practice (laboratory work and practical exercises)	I semester	English

Requirements for students	
Prerequisites: English B2 level; General genetics	**Additional requirements (if any):**

Course (module) volume in credits	Total student's workload	Contact hours	Self-study hours
10	260	120	140

Purpose of the course unit (module): programme competences to be developed		
The aim of the present course is to develop the abilities of student to understand the foundations of genomics and to apply knowledge of genomics to the analysis of the normal and pathological characteristics of a human and to the analysis of the personal genomics.		
Learning outcomes of the course unit (module)	**Teaching and learning methods**	**Assessment methods**
1. A fundamental understanding of the biology of human genome by describing genome structure and functional patterns at the molecular level and the mechanisms of physiological regulation	Lectures, debates, group discussion and practical assignments	Completion of practical assignments; Written project; written examination.
2. Be able to analyse, manage and model genomics data from the field of system biology	Lectures, debates, group discussion and practical assignments	
3. Be able to gather and analyse information on subjects related to system biology with a critical approach, and to carry out a technological watch	Lectures, debates, group discussion and practical assignments	
4. Be able to apply tools and techniques for exploring the personal genomics of ancestry and genealogy, discovery and interpretation of genetic trait associations	Lectures, debates, group discussion and practical assignments	
5. Perform duties within the deadlines and goals of a project	Practical assignments	
6. Be able to obtain information on digital representation of personal genomic data by performing practical and theoretical work in system biology in accordance with the bioethics requirements	Lectures, debates, group discussion and practical assignments	
7. Be able to identify the role of personal genomics in drug response by communicating in a clear manner with specialists from other fields or the public about professional project, on work results, or about the results of tasks	Lectures, debates, group discussion and practical assignments	
8. Be able to work autonomously and as a part of a multidisciplinary team; act honestly and according to ethical obligations by applying genomics knowledge in to practice	Lectures, debates, group discussion and practical assignments	
9. Be able to critically analyse their own research quantitative results and know possible ways for improvement	Lectures, debates, group discussion and practical assignments	

Content: breakdown of the topics	Contact hours							Self-study work: time and assignments	
	Lectures	Tutorials	Seminars	Exercises	Laboratory work	Internship/work placement	**Contact hours**	**Self-study hours**	**Assignments**
1. Genome structure and function. Main features of bacterial and viral genome. Genetic diversity mechanisms in prokaryotes. Main features of eukaryotes genomes. The architecture of human nuclear genome. Genes and gene-related sequences of nuclear genome. Protein-coding genes. RNA coding genes. Heterochromatin DNA and transposon repeats. Mitochondrial genome. Human Genome Project. Human gene expression. DNA and chromosome structure, copy number and function during the cell cycle. Cell division and transmission of DNA to daughter cells. Chromatin accessibility and conformation. Histones and other DNA binding proteins. Regulation by DNA methylation and noncoding RNAs. X inactivation, imprinting and epigenetic memory. RNA transcription and processing, regulation, post-transcriptional regulation. Translation, posttranslational processing and protein structure.	8			4			**12**	**14**	Preparation for seminar (self-study). Recommended reading: Lesk 2017, 1-32; 42-47; 179-231 Tom Strachan and Andrew Read 2018, 3-67; 203-242; 279-360
2. Genome databases and tools. Introduction to genome databases. Importance of databases in human genome analysis. Databases classification. Primary databases – GRC (Genome Reference Consortium), OMIM (Online Mendelian Inheritance in Man), Orphanet, ClinVar, GenBank, HapMap, 1000 Genomes Project, Protein Data Bank, GEO (Gene Expression Omnibus). Secondary databases - NCBI, Ensembl, UCSC. Main tools of Ensembl and UCSC and their use.				3			**3**	**4**	Preparation for laboratory work (self-study). https://www.ensembl.org/index.html https://www.ncbi.nlm.nih.gov/ https://genome.ucsc.edu/ Recommended reading: Lesk 2017, 104-109 Dong Zou et al., 2015
3. Technologies of genome structure and expression analysis. Cloning DNA in bacterial cells. Amplification of DNA by replication. Nucleic acid hybridization. Chain termination sequencing. Next generation sequencing. SNP-comparative genome hybridization, microarray comparative genome hybridization. Kariotyping. Fluorescent *in situ* hybridization.	13				11		**24**	**28**	Preparation for laboratory work (self-study). Recommended reading: Lesk 2017, 107-126 Tom Strachan and Andrew Read 2018, 161-202
4. Genetic variation between individuals. Variability of the human genome, molecular markers. Human genome variants classification. Single nucleotide variants and other point mutations (loss of function, gain of function, dynamic mutations, molecular pathology of mitochondrial genome). Mutations at cellular level. DNA repair	4				4		**8**	**9**	Preparation for seminar and laboratory work (self-study). Recommended reading:

systems. Chromosomal abnormalities and structural variants, microdeletions and microduplications. ISCN nomenclature. ACMG standards and guidelines for the interpretation of sequence variants.									Dudley and Karczewski 2013, 199-231 Tom Strachan and Andrew Read 2018, 361-418; 491-548
5. Genome evolution and comparative genomics. Human evolution. Main evolutionary forces, their definitions. Evolution of mammalian chromosomes. Evolution of genes and regulatory sequences, transposon origins of functional sequences. Main levels of evolution: gene, genomic, chromosome evolution. Sequence alignment, phylogeny: definitions, main methods. Concept, principle and benefits of the comparative genomics. Synteny. Orthologs and paralogs. Comparison of genomes between the same species, close related species, and very distinct species. Model organisms in Genomics research. Human origins. Human evolutionary history from genome sequences. Health consequences.			4	4			**8**	**9**	Preparation for seminar (self-study). Recommended reading: Lesk 2017, 143-178; 233-263; 295-328 Tom Strachan and Andrew Read 2018, 419-490
6. Genomics and human biology. Oncogenomics. Principles of cell signalling. Cell proliferation and programmed cell death. Cell adhesion and tissue formation. Immune system genomics. Cell lineages and tissue differentiation in early mammalian development. Stem cells and cell differentiation. Multistage cancer evolution, main mechanisms and functional pathways. Hereditary and sporadic cancer. Oncogenes and tumour suppressor genes – the mechanisms of their alterations and influence to cancer development. Cancer cells' genome and epigenome instability, genes implicated in maintenance of genome stability. Genetic markers in oncology, targeted treatment of cancer.	7						**7**	**8**	Preparation for seminar (self-study). Recommended reading: Lesk 2017, 218-294 Tom Strachan and Andrew Read 2018, 69-136; 597-630
7. Genome mapping and annotation. Next generation sequencing pipeline. Alignment to reference genome. Variant calling. Main parameters of next generation sequencing pipeline. Annotation. Annotation classifications.	4			12			**16**	**19**	Preparation for laboratory work (self-study). Recommended reading: Lesk 2017, 97-141
8. Clinical genomics. Monogenic and multifactorial inheritance. Mosaicism and new mutations. Non-Mendelian characters. Principles of genetic counselling. Mendelian pedigree patterns. Mendelian disorder risk of estimation to offspring. Mapping and identifying genes for monogenic disorders. Positional cloning. Haplotype sharing and autozygosity. Whole genome sequencing and whole exome sequencing based disease gene identification.	7		4				**11**	**13**	Preparation for seminar (self-study). Recommended reading: Tom Strachan and Andrew Read 2018, 137-160; 549-572 Murray, Babyatski, Giovanni 2014, 7-47
9. Genomics and the environment. Genome and environment interaction. Biological mechanisms. Epidemiological and molecular methods of analysis. Nutrigenomics. Toxicogenomics. Impact of metabiome to host physiology. Identification of susceptibility factors of complex disease and understanding pathogenesis. Epidemiological approaches. Linkage analysis. Whole genome	4			4			**8**	**9**	Preparation for seminar (self-study). Recommended reading: Dudley and Karczewski 2013, 163-180 Tom Strachan and Andrew Read 2018, 573-598

association studies in research of multifactorial disorders.									
10. Applied human genomics. The main challenges of personalized medicine. Clinical diagnostic testing. Population screening. DNA forensics. Genetic approaches to treating disease. Pharmacogenomics. Subject of Pharmacogenomics. Pharmacokinetics and Pharmacodynamics. Drug metabolism. Drug metabolism reactions. Enzymes involved in drug metabolism and their genetic diversity. Principles of genetic manipulation of mammalian cells. Gene therapy. Genome editing. Gene silencing. Germ-line transgenesis and transgenic animals.	4		4				**8**	**9**	Preparation for seminar (self-study). Recommended reading: Tom Strachan and Andrew Read 2018, 243-278; 631-666 Dudley and Karczewski 2013, 139-162
11. Systems biology. An understanding of systems biology and its applications. Systems biology and networks, types and properties. The metabolome networks. Regulatory networks, protein-protein, protein-nucleic acid interaction. Approaches and methodology in systems biology.	7		4	4			**15**	**18**	Preparation for seminar (self-study). Recommended reading: Lesk 2017, 443-482
Total	**56**		**14**	**35**	**15**		**120**	**140**	

Assessment strategy	**Weight,%**	**Deadline**	**Assessment criteria**
Control tests after each practical training or laboratory work (nine)	15 % (average mark of all the control tests)	During the semester	Evaluation method: written examination comprised of 3 short open-ended questions or tasks (0.5 mark each); detailed answer, the consistency and accuracy of the presented information.
Colloquia (two)	20 %	During the semester	Evaluation method: written examination comprised of 2 open-ended theoretical questions or tasks (0.5 mark each); detailed answer, the consistency and accuracy of the presented information.
Written project (one)	15 %*	During the semester	A comprehensive assessment of scientific publications.
Final examination: a test at the end of the course	50 %	December	Test consists of the 50 *closed-ended questions; each question* shall carry one mark, total marks being 50. Evaluation description: 10: Outstanding knowledge and skills. Evaluation level. 46-50 grades. 9: Good knowledge and skills with unessential mistakes. Synthesis level. 41-45 grades. 8: Average knowledge and skills with mistakes. Analysis level. 36-40 grades. 7-6: Below average knowledge and skills with substantial mistakes. Knowledge application level. Accordingly, 26-30 and 31-35 grades. 5: Knowledge and skills meet minimum criteria. Many mistakes. Level of knowledge and understanding. 21-25 grades. 4-0: Knowledge and skills do not meet minimum criteria. 20-0 grades.

* Additional 10 % for the excellence of the written project.

Author	**Year of publication**	**Title**	**Issue of a periodical or volume of a publication**	**Publishing place and house or web link**
Compulsary reading				
Tom Strachan and Andrew Read	2018	Human Molecular Genetics		5th ed., CRC Press
Arthur M. Lesk	2017	Introduction to genomics		3rd ed., Oxford
Joel T. Dudley and Konrad Karczewski	2013	Exploring personal genomics		Oxford

Michael F. Murray, Mark W. Babyatski, Monica A. Giovanni	2014	Clinical genomics. A practical applications in adult patient care		McGraw Hill ed.
Optional reading				
https://hstalks.com/biosci/				
Dong Zou, Lina Ma, Jun Yu, Zhang Zhang	2015	Biological databases for human research	13(1):55-63	Genomics Proteomics Bioinformatics

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