DESCRIPTION OF COURSE UNIT FOR DOCTORAL STUDIES AT VILNIUS UNIVERSITY

Scientific Area/eas, Field/ds of Science	Medical and Health Sciences (M 000), Medicine (M 001)			
Faculty, Institute, Department / Clinic	Faculty of Medicine, Institute of Biomedical Sciences, Department of Human and Medical Genetics			
Course unit title (ECTS credits, hours)	**Molecular Human Genetics and Genome Analysis** 10 credits (270 hours)			
Study method	**Lectures**	**Seminars**	**Consultation**	**Individual work**
Number of ECTS credits	**-**	**-**	1	9
Method of the assessment (in 10 point system)	Written and oral exam. Three open-ended/problem-based questions.			

PURPOSE OF THE COURSE UNIT

To learn and master the molecular aspects of the science of human genetics and the principles of genome structure and analysis.

THE MAIN TOPICS OF COURSE UNIT

The structure and function of DNA and RNA. The genetic code. The human genome: functional and regulatory elements, unique and repetitive sequences, mobile elements. Mechanisms of transposition. Mechanisms of formation of new genes by duplication. Catalogue of human genes. Chromosomes. Chromatin domains. Chromatin modifications. Methods of studying chromosomes. C, G, R, Q chromosome staining methods: principles and applications. Fluorescence in situ hybridisation. Comparative genomic hybridisation. Mechanisms and applications of DNA modification. DNA cloning: principles, tools and applications. Polymerase chain reaction, its modifications and applications. Cell cycle and its control. Mechanisms and theories of cancer development.

DNA damage repair: direct repair, excision repair, repair mechanisms for breaks, mispairing of bases in DNA. Mechanisms of homologous recombination. Mechanism and regulation of genome replication. Classification and nomenclature of mutations. Mechanisms of mutation generation: point mutations, dropouts, insertions, inversions, conversions, copy number changes. Analysis of new regulatory elements and DNA sequence fragments. Functional mechanisms of mutational effects. In silico assessment of the functional effects of DNA sequence changes. Molecular pathology: from disease to gene and from gene to disease. Methods for the identification of nucleotide sequence changes. Gene expression: mechanisms and regulation. Mechanisms of gene silencing. Molecular approaches to the study of gene expression. Gene expression studies using cell cultures and extracts. Ribosome structure. Mechanism and regulation of translation. iRNA maturation control. iRNA research methods.

Genome sequencing. DNA sequencing methods: their principles, advantages and disadvantages. RNA sequencing: methods and applications. Human Genome Project. Genome sequencing strategies. Evolution, tasks and main results of human genome research projects (1000 genomes, HapMap, ENCODE, LITGEN projects). Genome variant databases.

Mapping the human genome. Genetic mapping: markers, purpose and applications. Physical mapping: methods, advantages and disadvantages. Parametric and non-parametric linkage analysis. Non-parametric coupling. Strategies for mapping genomic regions of the genome that contribute to common complex diseases and traits. The principle of large-scale whole-genome analysis. Analysis and problems of the Lod estimate. Disease-associated gene mapping depending on gene location: positional cloning, functional cloning. Confirmation/validation of candidate genes.

Methods and applications of transcriptome research. Methods and applications of proteome research. DNA-protein interactions. Motifs of DNA-binding proteins. Methods for the identification of genetic regions that bind proteins. Methods for the detection of DNA-protein interactions. Identification of regulatory sequences using reporter genes and DNA-protein interactions.

Genome editing: genome editing technologies. CRISPR/Cas system principle and applications. Gene therapy: strategies, principles, advantages and disadvantages.

Comparative genomics. Intron evolution, possible mechanisms of origin. Principles of phylogenetic tree construction based on reconstructed DNA and applications. Animal models for the study of changes in the human genome. Transgenic animals, their applications. Applications of embryonic stem cells. Regulation and bioethical, social and legal issues concerning the research and manipulation of the human genome.

RECOMMENDED LITERATURE SOURCES

1. Tom Strachan, Andrew Read, Human Molecular Genetics, 5th. ed., 2019.
2. Guy Bradley-Smith, Sally Hope, Helen V.Firth, Jane A.Hurst, Oxford Handbook of Genetics, (Oxford Handbooks Series), Oxford university press, 2009.
3. Speicher, Michael; Antonarakis, Stylianos E.; Motulsky, Arno G. (Eds.). Vogel and Motulsky's Human Genetics. Problems and Approaches,4th. ed., Springer,2010.
4. Daniel L. Hartl and Andrew G. Clark Principles of Population Genetics, Fourth Edition. Sinauer Associates, Inc. Publishers. 2007.
5. Barnes MR and Gray IC. Bioinformatics for geneticists. Wiley, 2nd ed, 2007.
6. http://varnomen.hgvs.org/
7. https://genome.ucsc.edu/
8. http://ensmebl.org/

CONSULTING LECTURERS

1. Coordinating lecturer: Laima Ambrozaitytė (Assoc. Prof. Dr.).
2. Tautvydas Rančelis (Dr.).
3. Ingrida Domarkienė (Assist. Prof. Dr.).

APPROVED:

By Council of Doctoral School of Medicine and Health Sciences at Vilnius University: 29th of September 2022

Chairperson of the Board: Prof. Janina Tutkuvienė