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); Public Health (M 004) Natural Sciences (N 000): Biology (N 010)			
Faculty, Institute, Department/Clinic	Faculty of Medicine Institute of Biomedical Sciences Department of Human and Medical Genetics			
Course unit title (ECTS credits, hours)	**Population Genetics and Genomics** 9 credits (243 hours)			
Study method	Lectures	Seminars	Consultations	Self-study
Number of ECTS credits	-	-	2	7
Method of the assessment (in 10 point system)	Exam: oral consisting of three open questions.			

PURPOSE OF THE COURSE UNIT

To provide a basic fundamental knowledge on human population and evolutionary genetics.

THE MAIN TOPICS OF COURSE UNIT

Main part: The scope of population genetics. Exponential and logistic population growth models.

Human Genome Variation. Allele, genotype and the phenotype. Genotype and allele frequencies in population. Genetic markers used to analyse genetic variation: single nucleotide polymorphisms, mtDNA, variation in tandemly repeated DNA sequences, transposable elements, structural variation, pros and cons.

Methods used to detect genome diversity. The polymerase chain reaction (PCR), Sanger sequencing, the human reference sequence, next-generation sequencing, high throughput SNP chips, phasing and methods used to determine haplotypes, databases of sequence variation.

Processes shaping genome diversity. Hardy-Weinberg equilibrium, evolution and its applications: heterozygote frequency, Hardy-Weinberg equilibrium and dominant alleles, more than two alleles, X-linked genes. Deviations from Hardy-Weinberg equilibrium. Linkage disequilibrium and equilibrium. Inbreeding coefficient, types of inbreeding. The impact of inbreeding on genotype frequencies. Genetic impact of inbreeding in human populations. Types of mutation and their evolutionary impact, fixation of new mutations in population, neutral mutations, reversible and irreversible mutation models, changes in allele frequency in population. New combinations of alleles during meiosis recombination, impact of recombination on evolution. Genetic drift: binomial sampling, Wright and Fisher gene drift model, effective population size, different parts of the genome, changes in effective population size, population subdivision and effective population size, variation in census population size and influence of reproductive success on effective population size, relation between genetic drift and population size variation, genetic drift and inbreeding, genetic drift in isolated populations, coalescent theory and demographic history. Natural selection: absolute and relative fitness, a general model of natural selection, types of natural selection, selection and inbreeding, natural selection in human populations, haemoglobin S and malaria, the Duffy blood groups and malaria, the evolution of human skin colour. Migration and gene flow: one-way migration "the island model", two-way gene flow, gene flow interaction with genetic drift, admixture model. Equilibrium between evolutionary factors: equilibrium between mutations

and genetic drift, between recombination and genetic drift, mutations and natural selection. The neutral theory of molecular evolution: the molecular clock.

Genetic diversity in population analysis. Average heterozygosity, heterozygosity – measure of genetic diversity, genetic distances between populations using Fst or Nei's D statistics, calculation of distances between alleles using models of mutation, calculation of genetic distances between individuals using genome-wide data, population structure analysis using genome data, isolate breaking, Wahlund effect. Phylogenetics: types of phylogenetic trees and construction. Origins of modern humans. Evidence from fossils and morphology, evidence form archaeology and linguistics, hypothesis to explain the origin of modern humans, evidence from the genetics of present-day populations, evidence from ancient DNA.

The computational methods in population genomics. The data clustering and visualization methods. The basic dimension reduction methods of multidimensional data: multidimensional scaling, principal component analysis. Apps for data clustering and visualization overview (R, Matlab, Past, SPSS).

RECOMMENDED LITERATURE SOURCES

1. Daniel L. Hartl. (2020) A Primer of Population Genetics and Genomics. Oxford Scholarship Online.
2. Tom Strachan, Andrew Read. (2018) Human Molecular Genetics (5th edition). 784 p., Garland Science.
3. Jobling M., Hollox E., Hurles M., Kivisild T., Tyler-Smith C. (2014) Human Evolutionary Genetics (2nd edition). 670p., Garland Science, New York/London.
4. Relethford J. H. (2012) Human Population Genetics (1st edition). 316 p., Wiley-Blackwell.
5. Kučinskas V. (2004) Genomo įvairovė: lietuviai Europoje. 287 p., Spalvų šalis, Vilnius.
6. Kučinskas V. (1994) Įvadas į Žmogaus populiacinę genetiką. Vilniaus universiteto leidykla, Vilnius.
7. Molytė A, Kučinskas V. (2015). Žmogaus genomo analizė: dažniausiai taikomi matematiniai metodai. 96 p., Vilniaus universiteto leidykla, Vilnius.

CONSULTING LECTURERS

1. Coordinating lecturer: Alina Urnikytė (Dr.)
2. Vaidutis Kučinskas (Acad.Prof. Habil. Dr.).
3. Alma Molytė (Assoc. 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