

SYLLABUS

Winter semester 2022/2023

(Lectures will be held on Wednesdays starting on 26 October 2022 in lecture hall of the Institute of Human Genetics Polish Academy of Sciences)

Course	Human Genome Evolution and Variability in Health and Disease
Host Institution	Institute of Human Genetics, Polish Academy of Sciences Strzeszyńska Street, 32
Language	English
The expected effects of teaching in terms of: knowledge, skills and social qualifications	Ph. D. student is supposed to: 1. Gain knowledge in several aspects of human genome evolution and variability and its significance in human health and disease. In particular, the following specific topics will be elaborated: -Mechanisms shaping the genetic variability -Evolution of sex chromosomes -Selection and demography in shaping genetic diversity profiles -Evolution of <i>Homo sapiens</i> in light of the genetic studies -Copy number variation in the human genome -Mobile elements in the human genome -Regulatory noncoding sequences in the human genome -Mitochondrial haplogroups -Genetic epidemiology in forensic studies -Genetic epidemiology in medical studies -Genetic variability of the cell function in health and disease 2. To get familiar with new directions in the field of studying genetic variability in the context of human diseases 3. To learn how to formulate scientific questions related to human genome evolution and variability in health and disease
Type of course	Obligatory
Semester/year	Winter semester 2022/2023

First name/family name of the person responsible for the course	Prof. dr hab. Ewa Ziętkiewicz
First name/family name of the person responsible for the exam	Prof. dr hab. Ewa Ziętkiewicz
Format	Lecture will be held in English with usage of audio-visual equipment. Lecture will be followed by discussion
Basic and additional requirements	Skills in English and knowledge in molecular biology
Number of ECTSs	3 ECTS
ECTSs summary	1 ECTS corresponds to 25-30 hours of personal studies focused on broadening knowledge based on suggested bibliography (<i>vide</i> bibliography list below).
Method of teaching	Lectures will be held using PowerPoint presentation and a multimedia projector
Method of evaluation	Written exam
Prerequisite for passing	Positive score at the exam
Topics	<ul style="list-style-type: none"> -Mechanisms how genetic variability arises and how it spreads within populations -Evolution of sex chromosomes -Role of selection in creating genetic diversity profiles -Evolution of Homo sapiens in the context of genetic diversity in the modern human populations -Diversity of repetitive elements within the genome (copy number variation, mobile elements) -Regulatory noncoding sequences in the human genome -Practical applications of genomic diversity -Genetic epidemiology in medical and forensic studies -Genetic bases of the variability of the cell function in health and disease
Additional material	<p>Presentation of each lecture in PDF format</p> <p>Bibliography related to each lecture</p>

Bibliography

LL Cavalli-Sforza & Bodmer The Genetics of Human Populations. Dover Publications (2013)

ES Tobias, M Connor & M Ferguson-Smith. Essential Medical Genetics Essential Medical Genetics. Wiley-Blackwell (2011).

DL Hartl & AG Clark. Principles of population genetics. Sinauer Associates (2006)

KR Veeramah & MF Hammer. The impact of whole-genome sequencing on the reconstruction of human population history. *Nature Reviews | Genetics* 15:149-161 (2014).

S Eggers & A Sinclair. Mammalian sex determination—insights from humans and mice. *Chromosome Research* 20:215–238 (2012).

BJ Lesch and DC Page. Genetics of germ cell development. *Nature Reviews Genetics* 13:781-794 (2012).

N Naidoo, Y Pawitan, R Soong, DN Cooper and CS Ku. Human genetics and genomics a decade after the release of the draft sequence of the human genome. *Human Genomics*. 5:577–622 (2011).

MM Matzuk & DJ Lamb. The biology of infertility: research advances and clinical challenges. *Nature Medicine* 14:1197-1213 (2008).

N Powles-Glover. Cilia and ciliopathies: Classic examples linking phenotype and genotype: An overview. *Reproductive Toxicology* 48:98–105 (2014).

Zietkiewicz, Witt, Daca, Żebracka-Gala, Goniewicz, Jarzab & Witt. Current genetic methodologies in the identification of disaster victims and in forensic analysis. *J Appl Genetics* 53:41–60 (2012).