

SYLABUS

Winter semester 2026/2027

(Lectures will be held Wednesdays starting on 10 October 2026 in lecture hall of the Institute of Human Genetics Polish Academy of Sciences)

Course	Human Genome Variability in Health and Disease
Host Institution	Institute of Human Genetics 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 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 -History of genes and history of populations -Mobile elements in the human genome -Evolution of Homo sapiens -Genetic epidemiology in medical studies -Genetic variability of the cell function in health and disease -human intestinal microbiome 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 variability in health and disease
Learning outcomes, according to Attachment 2 to the Programme of the Study (valid from October 1 st 2026)	P8S_WG (pkt. 1, 2 and 3) P8S_UW (pkt. 1 and 2) P8S_UK (pkt. 3)
Type of course	Obligatory for medical sciences

Semester/year	Winter semester 2026/2027
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 power point presentation and a multimedia projector
Method of evaluation	Written exam
Prerequisite for passing	Positive score at the exam
Topics	-Mechanisms how genetic variability arises and how it spreads within populations -Evolution of sex chromosomes -History of genes and history of populations -Role of selection in creating genetic diversity profiles -Diversity of the human genome -Diversity of repetitive elements within the genome -Evolution of <i>Homo sapiens</i> in the context of genetic diversity in the modern human populations -Practical applications of genomic diversity -Some aspects of genetic bases of functional modulation of the cell in health and disease -Mechanisms and factors shaping the genetic variability -Evolution of sex chromosomes -History of genes and history of populations -Role of selection and demography in shaping genetic diversity profiles -Evolution of the human genome -Repetitive elements in the human genome -Evolution of <i>Homo sapiens</i> in light of the genetic diversity in modern human populations

	-Genetic epidemiology in medical and forensic studies -Genetic bases of the variability of the cell function in health and disease -human intestinal microbiome in health and disease
Additional material	Presentation of each lecture in PDF format Bibliography related to each lecture
Bibliography	LL Cavalli-Sforza & Bodmer The Genetics of Human Populations. Dover Publications (2013) ES Tobias, M Connor & M Ferguson-Smith. 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).