University of Gdańsk

Subject card

Subject name and code	Human Genome Portrait, PG_00209703		
Field of study	Biology		
Date of commencement of studies	October 2024	Academic year of realisation of subject	
Education level	Bachelor's studies	Subject group	Obligatory subject group in the field of study Optional subject group
Mode of study	full-time studies	Mode of delivery	e-learning
Year of study	3	Language of instruction	English The course is delivered fully online. Schedule and task deadlines are provided on eg. the eLearning platform. The course does not include medical diagnosis or interpretation of students' private genetic test results.
Semester of study	6	ECTS credits	2.0
Learning profile	academic	Assessment form	credit
Conducting unit			
Name and surname of lecturer (lecturers)	Subject supervisor	prof. dr hab. Magdalena Gabig-Cimińska	
	Teachers		

Lesson types

Lesson type	Lecture	Tutorial	Laboratory	Project	Seminar	SUM
Number of study hours	30.0	0.0	0.0	0.0	0.0	30

E-learning hours included: 30.0

Learning activity and number of study hours

Learning activity	Participation in didactic classes included in study plan	Participation in consultation hours	Self-study	SUM
Number of study hours	30	0.0	25.0	55

Subject objectives

The aim of the course is to present the human genome as a dynamic biological "portrait" of an individual and of populations: its organisation, variability, methods of reading and interpretation, and relevance to health, ancestry and human traits. Students learn the foundations of human genomics, sequencing and genotyping technologies, the use of key genomic databases, and the limitations of variant interpretation. A major objective is to develop the ability to critically evaluate personal, population and medical genomics applications while considering ethical, legal and social implications.

Learning outcomes	Course outcome	Subject outcome	Method of verification
		The student describes the organisation of the human genome, coding and non-coding sequence classes, and the main types of genomic variation: SNPs, indels, CNVs, STRs, structural variants and mtDNA variation. The student explains the meaning of reference genomes, pangenomes and population datasets, and understands limitations related to reference population choice, data quality and algorithmic bias. The student retrieves and interprets basic information on a gene or variant using resources such as NCBI, Ensembl, UCSC Genome Browser, gnomAD, ClinVar, GWAS Catalog and IGSR. The student critically evaluates example personal genomics reports, distinguishes descriptive information from clinical conclusions, and formulates cautious conclusions with stated limitations. The student identifies ethical, legal and social consequences of working with genomic data, including privacy, consent, discrimination, secondary findings and responsible risk communication.	[SW4] test/exam - oral or written [SU1] oral statement/conversation/ discussion [SU4] test/exam - oral or written [SU8] observation of student's independent or team work [SK1] oral statement/conversation/ discussion [SK4] test/exam - oral or written [SK8] observation of student's independent or team work
Subject contents	1. The human genome as a biological “portrait”: from the Human Genome Project to reference genomes and pangenomes. 2. Genome organisation: chromosomes, genes, regulatory regions, repetitive sequences, the mitochondrial genome and mobile elements. 3. Genomic variation: SNPs, indels, CNVs, STRs, structural variants, haplotypes, linkage and population frequencies. 4. Genome analysis technologies: genotyping, Sanger sequencing, next-generation sequencing, long-read sequencing and basic data quality control. 5. Variant annotation and interpretation: functional consequences, allele frequency, pathogenicity, interpretative uncertainty and ACMG/AMP classification. 6. Data resources: NCBI, Ensembl, UCSC Genome Browser, dbSNP, ClinVar, gnomAD, 1000 Genomes/ IGSR, GWAS Catalog and OMIM. 7. Personal and medical genomics: direct-to-consumer tests, disease risk, pharmacogenomics, secondary findings and polygenic risk scores. 8. Population genomics: ancestry, migrations, natural selection, diversity and limits of ancestry inference. 9. The epigenome, transcriptome and gene-expression regulation as extensions of the human genomic portrait. 10. ELSI aspects: data privacy, informed consent, data sharing, security, equity and responsible communication of results.		
Prerequisites and co-requisites	Basic knowledge of general and molecular genetics, cell biology and human biology. Basic English-language scientific literacy and the ability to use simple web-based tools are useful.		

<table>
<tr><td>Assessment methods and criteria</td><td colspan="2"><table>
<tr><th>Subject passing criteria</th><th>Passing threshold</th><th>Percentage of the final grade</th></tr>
<tr><td>Course activity: short problem-based tasks, online discussion participation and timely completion of modules.</td><td>51.0%</td><td>20.0%</td></tr>
<tr><td>Tests and quizzes on eg. the eLearning platform verifying knowledge after thematic modules.</td><td>51.0%</td><td>80.0%</td></tr>
</table></td></tr>
<tr><td rowspan="3">Recommended reading</td><td>Basic literature</td><td>• Strachan T., Read A. P. (2018). Human Molecular Genetics. 5th ed. Garland Science / CRC Press.
• Brown T. A. (2023). Genomes 5. 5th ed. Garland Science / CRC Press.
• Human Pangenome Reference Consortium. (2023). A draft human pangenome reference. Nature, 617, 312–324. DOI: 10.1038/s41586-023-05896-x.</td></tr>
<tr><td>Supplementary literature</td><td>• Richards S. et al. (2015). Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the ACMG and AMP. Genetics in Medicine, 17, 405–424. DOI: 10.1038/gim.2015.30.
• Lee K. et al. (2025). ACMG SF v3.3 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the ACMG. Genetics in Medicine, 27(8), 101454. DOI: 10.1016/j.gim.2025.101454.
• 1000 Genomes Project Consortium. (2015). A global reference for human genetic variation. Nature, 526, 68–74. DOI: 10.1038/nature15393.
• Karczewski K. J. et al. (2020). The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 581, 434–443. DOI: 10.1038/s41586-020-2308-7.</td></tr>
<tr><td>eResources addresses</td><td>Basic
https://www.internationalgenome.org/ - 1000 Genomes / IGSR
https://www.ebi.ac.uk/gwas/ - GWAS Catalog
https://www.ncbi.nlm.nih.gov/clinvar/ - ClinVar
https://gnomad.broadinstitute.org/ - gnomAD
https://genome.ucsc.edu/ - UCSC Genome Browser
https://www.ensembl.org/Homo_sapiens/Info/Index - Ensembl Human
https://www.ncbi.nlm.nih.gov/genome/guide/human/ - NCBI Genome / Human genome resources
Supplementary
https://www.ncbi.nlm.nih.gov/snp/ - dbSNP
https://www.omim.org/ - OMIM
https://www.genome.gov/ - NHGRI – genomics and policy resources
https://humanpangenome.org/ - Human Pangenome Reference Consortium</td></tr>
<tr><td>Example issues/ example questions/ tasks being completed</td><td colspan="2">1. Explain the difference between a reference genome and a pangenome. What are the limitations of a single reference genome?

2. For a selected variant, read the allele frequency in chosen populations in gnomAD and propose a cautious interpretation.

3. Compare information about the same gene in Ensembl, NCBI and the UCSC Genome Browser. What types of data are available?

4. Using a ClinVar record, identify which elements support or limit interpretation of variant pathogenicity.

5. Critically assess an excerpt from a personal genomics test report: what is descriptive, what requires validation, and what cannot be inferred?

6. Discuss key risks to privacy and autonomy when a person’s genomic data are analysed or shared.</td></tr>
<tr><td>Work placement</td><td colspan="2">Not applicable</td></tr>
</table>

Document generated electronically. Does not require a seal or signature.