

Subject card

Subject name and code	Genetics and human genetic diseases, PG_00156415						
Field of study	Bioinformatics						
Date of commencement of studies	October 2024		Academic year of realisation of subject		2026/2027		
Education level	Bachelor's studies		Subject group		Optional subject group		
Mode of study	full-time studies		Mode of delivery		at the university		
Year of study	3		Language of instruction		Polish		
Semester of study	6		ECTS credits		3.0		
Learning profile	academic		Assessment form		credit		
Conducting unit	Pracownia Molekularnych i Komórkowych Podstaw Strategii Nutr -> Department of Medical Biology and Genetics -> Faculty of Biology -> Rector						
Name and surname of lecturer (lecturers)	Subject supervisor		dr Marta Moskot				
	Teachers						
Lesson types	Lesson type	Lecture	Tutorial	Laboratory	Project	Seminar	SUM
	Number of study hours	0.0	0.0	30.0	0.0	0.0	30
	E-learning hours included: 0.0						
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		45.0	75
Subject objectives	Getting to know the human karyotype with particular emphasis on the location of genes whose mutations are associated with hereditary diseases, disorders of the pattern of inheritance of Mendelian traits, polygenic traits and epigenetic control of gene expression in molecular diagnostics to assess the risk of human genetically determined diseases, and with methods of studying the human genome in forensic medicine and forensics.						
Learning outcomes	Course outcome		Subject outcome		Method of verification		
	[BIOINL3_K03] Has an awareness and understanding of the risks and dilemmas, including ethical dilemmas, involved in conducting scientific research and introducing advanced technologies; understands and appreciates the importance of intellectual property; acts ethically		Student is able to analyze scientific publications in the field of human genetics in terms of their use in formulating and solving problems related to bioinformatics. Can critically analyze information about human genetics from various available sources.		[SK1] oral statement/conversation/discussion		
	[BIOINL3_W02] Has the scientific knowledge necessary to understand the basic processes in living organisms.		Student has in-depth knowledge of human genetics. Knows the molecular mechanisms of genetic information transfer and gene expression. Knows the genetic basis of human diseases.		[SW1] oral statement/conversation/discussion [SW2] presentation/project/paper/report [SW3] text preparation/written work		
	[BIOINL3_U02] Graduate is able to apply knowledge of natural sciences and science to formulate, analyze and solve problems related to bioinformatics		Student understands current problems related to bioethics. She knows the legal, organizational and ethical conditions of conducting human genetic research. He analyzes knowledge in the field of genetics and is able to indicate its practical applications in human life.		[SU1] oral statement/conversation/discussion [SU2] presentation/project/paper/report [SU3] text preparation/written work		

Subject contents	Getting to know the human karyotype with particular emphasis on the location of genes whose mutations are associated with hereditary diseases, disorders of the pattern of inheritance of Mendelian traits, polygenic traits and epigenetic control of gene expression in molecular diagnostics to assess the risk of human genetically determined diseases, and with methods of studying the human genome in forensic medicine and forensics.		
Prerequisites and co-requisites	After completing compulsory subjects in the first three semesters, the student has the knowledge and skills that qualify him or her to participate in and pass the course.		
Assessment methods and criteria	Subject passing criteria	Passing threshold	Percentage of the final grade
	Tasks	51.0%	40.0%
	Colloquium	51.0%	60.0%
Recommended reading	Basic literature	Friedman J.M. Dill F.J. Hayden M.R. McGillivray B.C. Genetyka Urban and Partner, 2000	
	Supplementary literature	Bradley J.T. Johnson D.R. Pober B.R. Genetyka medyczna. Wydawnictwo Lekarskie PZWL, 2008	
	eResources addresses		
Example issues/ example questions/ tasks being completed	1. Structure of the human karyotype characteristics of successive chromosomes.2. Genetic polymorphism.3. Disorders of the pattern of inheritance of Mendelian traits.4. Polygenic inheritance.5. Epigenetic control of gene expression.6. Teratogenesis and mutagenesis.7. Classification of human genetic diseases.8. Overview of single-gene diseases - gene background, symptoms, treatment.9. The process of lyonization and disorders of this process diseases linked to the X chromosome.10. Degeneration of the Y chromosome and its consequences disorders associated with mutations of the Y4 chromosome.11. Mitochondrial diseases.12. Prenatal and pre-implantation diagnostics.13. Diagnostics of genetic diseases - the use of molecular methods in the diagnosis of genetically determined diseases.14. Molecular diagnostics in forensic medicine and forensic science.15. Genetic counseling.		
Work placement	Not applicable		

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