University of Gdańsk

Subject card

Subject name and code	Human genetic disorders, PG_00147774		
Field of study	Genetics and Experimental Biology		
Date of commencement of studies	October 2024	Academic year of realisation of subject	2026/2027
Education level	undergraduate studies	Subject group	Obligatory subject group in the field of study 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	5	ECTS credits	1.0
Learning profile	academic	Assessment form	
Conducting unit	Pracownia Genomiki i Genetyki Człowieka -> Katedra Biologii i Genetyki Medycznej -> Faculty of Biology		
Name and surname of lecturer (lecturers)	Subject supervisor	dr hab. Joanna Jakóbkiewicz-Banecka	
	Teachers		

Lesson types

Lesson type	Lecture	Tutorial	Laboratory	Project	Seminar	SUM
Number of study hours	0.0	15.0	0.0	0.0	0.0	15

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	15	2.0	8.0	25

Subject objectives

1. Familiarization with the molecular basis of exemplary hereditary and cancerous diseases, as well as the main directions in the treatment of genetically determined diseases.
2. Understanding examples of single-gene and multi-gene diseases in clinical, molecular, and diagnostic aspects.
3. Acquaintance with the structure of human chromosomes, with particular emphasis on genes whose mutations are responsible for the occurrence of hereditary diseases.
4. Understanding and characterizing phenomena such as lyonization and its disorders, and Y chromosome degeneration.

Learning outcomes

Course outcome	Subject outcome	Method of verification
[GBEL3_U04] Capable of reading scientific texts in English and Polish with comprehension, synthesizing the knowledge contained within them, preparing well-documented studies on biological issues, as well as those related to research commercialization.	Is able to read and comprehend scientific texts in both English and Polish, synthesize the knowledge contained in them, and prepare well-documented studies on biological issues.	[SU1] oral statement/conversation/ discussion [SU4] test/exam - oral or written
[GBEL3_U09] Plan and pursue one's education autonomously and in a focused manner.	Is able to plan their education and learn independently and purposefully.	[SU4] test/exam - oral or written
[GBEL3_K04] Application of the principles of bioethics.	Consciously applies the principles of bioethics.	[SK4] test/exam - oral or written [SK8] observation of student's independent or team work
[GBEL3_K06] Integrity and honesty in scientific and professional work.	Understands the need for honesty and integrity in scientific and professional work.	[SK4] test/exam - oral or written [SK8] observation of student's independent or team work
[GBEL3_W03] The molecular mechanisms of genetic information transmission and gene expression, as well as the molecular and genetic basis of human physiology and diseases, including infectious diseases.	Understands the molecular mechanisms of genetic information transfer and gene expression, as well as the molecular and genetic basis of human diseases.	[SW4] test/exam - oral or written
[GBEL3_W06] the development and current state of knowledge, as well as the latest trends in molecular genetics and related fields; indicating their relationship with other disciplines in the natural or medical sciences and the possibilities of their practical application.	Is knowledgeable about the development and current state of knowledge as well as the latest trends in molecular genetics; identifies their connection with other natural or medical science disciplines and their potential practical applications.	[SW4] test/exam - oral or written
[GBEL3_W11] legal, organizational, and ethical considerations in conducting and implementing research in the field of genetics and experimental biology.	Is familiar with the legal, organizational, and ethical conditions for conducting and implementing research in the field of molecular genetics.	[SW4] test/exam - oral or written
[GBEL3_K07] Lifelong learning and updating knowledge in the field of molecular genetics and other disciplines.	Understands the need for continuous learning and updating knowledge in the field of molecular genetics and other related disciplines.	[SK1] oral statement/conversation/ discussion [SK4] test/exam - oral or written

Subject contents

- Classification of Human Genetic Diseases
- Multifactorial diseases
- The process of lyonization and its disorders X-linked diseases
- Degeneration of the Y chromosome and its consequences disorders associated with Y chromosome mutations
- Structure of the human karyotype characterization of individual chromosomes
- Comprehensive overview of single-gene diseases genetic basis, symptoms, treatment

Prerequisites and co-requisites

Scope of knowledge in genetics, including human genetics.

Assessment methods and criteria

Subject passing criteria	Passing threshold	Percentage of the final grade
test exam	51.0%	100.0%

Recommended reading

Basic literature	J.M Friedman, F.J.Dill, M.R. Hayden, B.C. McGillivray: Genetyka. (red. wyd. pol. J. Limon), Urban & Partner Genetyka medyczna, L.B. Jorde, J.C. Carey, M.J. Bamshad,, red. M. Borowiec, Edra Urban & Partner, 2021 J.M. Connor, M.A. Ferguson-Smith: Podstawy genetyki medycznej. PZWL
Supplementary literature	EPIGENETYKA, John C. Lucchesi, PWN, 2021 GENETYKA MEDYCZNA I MOLEKULARNA, Jerzy Bal, PWN, 2017 GENETYKA MEDYCZNA, Bogdan Kałużewski , Casey Carey , Lynn Jorde , Michael J. Bamshad, Edra Urban & Partner, 2013 GENETYKA MEDYCZNA, Edward Tobias, PZWL, 2014
eResources addresses	Adresy na platformie eNauczanie:

Example issues/ example questions/ tasks being completed

Work placement

Not applicable

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