University of Gdańsk

Subject card

Subject name and code	Human genetic disorders, PG_00154168		
Field of study	Biology		
Date of commencement of studies	October 2023	Academic year of realisation of subject	2024/2025
Education level	postgraduate 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	2	Language of instruction	Polish
Semester of study	3	ECTS credits	1.0
Learning profile	academic	Assessment form	
Conducting unit	Pracownia Genomiki i Genetyki Człowieka -> Katedra Biologii i Genetyki Medycznej -> Faculty of Biology -> Rektor		
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	15.0	0.0	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

- Familiarizing oneself with the molecular basis of exemplary hereditary and cancerous diseases; with the main directions in the treatment of genetically determined diseases.
- Learning about examples of single-gene and multi-gene diseases - in clinical, molecular, and diagnostic aspects.
- Getting acquainted with the structure of human chromosomes, with particular emphasis on genes whose mutations are responsible for the occurrence of hereditary diseases.
- Learning and characterizing phenomena such as lyonization and its disorders, and Y chromosome degeneration.

Learning outcomes

Course outcome	Subject outcome	Method of verification
[BIOLMU2_W05] the graduate understands the dynamic development of the biological sciences and is familiar with new research directions and disciplines	Possesses knowledge in the currently discussed issues related to the application of biological achievements in the molecular diagnosis of diseases.	[SW4] test/exam - oral or written
[BIOLMU2_K07] the systematic updating of biological knowledge and information on its practical applications	Analyzes knowledge in the fields of genetics, molecular diagnostics, and genetically determined diseases, and is able to identify its practical applications.	[SK4] test/exam - oral or written
[BIOLMU2_K04] correctly identify and resolve dilemmas related to the profession of biologist	Correctly identifies and resolves dilemmas related to the practice of the biologist profession.	[SK4] test/exam - oral or written
[BIOLMU2_U02] be fluent in the scientific literature of the biological speciality studied	Proficiently utilizes scientific literature in the field of human genetic diseases.	[SU4] test/exam - oral or written
BIOLMU2_W04	Recognizes the dynamic development of biological sciences in areas such as genomics and personalized medicine based on genetic knowledge.	[SW4] test/exam - oral or written
BIOLMU2_U07	Critically confronts information from various available sources and draws justified conclusions based on this.	[SU1] oral statement/conversation/ discussion [SU4] test/exam - oral or written

Subject contents

- The process of lyonization and its disorders, sex-linked diseases.
- Degeneration of the Y chromosome and its consequences, as well as diseases associated with it.
- Structure of the human karyotype - characterization of individual chromosomes.
- Comprehensive overview of single-gene diseases - genetic basis, symptoms, treatment.

Prerequisites and co-requisites

Basics of Human Genetics - knowledge of the specifics of the human genome and methods used in human genetics; understanding the basics of genetic disorders.

Human Genetics - knowledge about deviations from monogenic inheritance; understanding factors that complicate inheritance patterns; ability to identify the application of genetic methods in practice.

Assessment methods and criteria

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

Recommended reading

Basic literature

1. Jorde LB, Carey JC, Bamshad MJ, Genetyka medyczna, red. wyd. polskiego Maciej Borowiec, wydanie 6, Edra Urban&Partner, 2021.

2. Genetyka medyczna i molekularna, red. J. Bal, Warszawa, Wydawnictwo Naukowe PWN, 2017.

3. Tobias E. S., Connor M., Ferguson-Smith M., Genetyka medyczna, red. wyd. pol. A. Latos-Bieleńska, Warszawa, PZWL Wydawnictwo Lekarskie, 2013.

4. Lucchesi JC. Epigenetyka. PWN, Warszawa, 2022.

5. Drewa G, Ferenc T. Genetyka medyczna, Wrocław, 2011.

	Supplementary literature	1. Fletcher H, Hickey I, Krótkie wykłady: Genetyka, PZWL 2021. 2. Węgleński P. Genetyka molekularna, wydanie VI, PWN, 2020. 3. Genetyka kliniczna nowotworów, red. J. Lubiński, Szczecin, Print Group, 2018. 4. Medycyna personalizowana, red. A. Fronczak, Warszawa, Oficyna Wydawnicza Warszawskiego Uniwersytetu Medycznego, 2016. 5. Brown T. A., Genomy, Warszawa, Wydawnictwo Naukowe PWN, 2019.
	eResources addresses	Adresy na platformie eNauczanie:
Example issues/ example questions/ tasks being completed		
Work placement	Not applicable	

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