

## SYLABUS

Concerning the cycle of education **2024-2030**

Academic year 2025/2026

### 1. BASIC INFORMATION CONCERNING THIS SUBJECT

Subject	General genetics
Course code *	Gen/C
Faculty of (name of the leading direction)	Faculty of Medicine
Department Name	Department of General Genetics, Department of General and Clinical Genetics, Department of Biology
Field of study	Medicine
level of education	Uniform master's studies
Profile	General academic
Form of study	Stationary/ non- stationary
Year and semester	Year II, semester IV
Type of course	Obligatory
Language	English
Coordinator	Assoc. Prof. Izabela Zawlik
First and Last Name of the Teachers	Assoc. Prof. Izabela Zawlik Paweł Wołkow, PhD Aleksander Myszka, PhD

\* - According to the resolutions of Educational Unit

#### 1.1. Forms of classes, number of hours and ECTS

Semester No.	Lecture	Exercise	Conversation	Laboratory	Seminar	Z P	Praktical	Other	Number of points ECTS
IV	20	10	-	-	-	-	-	-	2

#### 1.2. The form of class activities

☒ classes are in the traditional form

☐ classes are implemented using methods and techniques of distance learning

#### 1.3. Examination Forms (exam, credit with grade or credit without grade)

### 2. BASIC REQUIREMENTS

The student should know the basics of biochemistry.

### 3. OBJECTIVES, OUTCOMES, AND PROGRAM CONTENT USED IN TEACHING METHODS

#### 3.1. Objectives of this course

C1	Getting to know basic terms and the most important discoveries in the field of genetics
C2	Understanding the impact of genes on cellular processes and phenotype
C3	Understanding the principles of inheritance of monogenic and multifactorial features
C4	Understanding the types and effects of chromosomal aberrations
C5	Understanding the diagnostic possibilities of diseases conditioned by chromosomal aberrations
C6	Understanding the diagnostic possibilities of diseases caused by gene mutations
C7	Acquiring the ability to recognize the way inheritance of human traits and diseases
C8	Acquiring the ability to select appropriate genetic tests depending on the type of health problems
C9	Acquiring the ability to interpret the results of cytogenetic and molecular research

#### 3.2. Outcomes for the course

EK (the effect of education)	The content of learning outcomes defined for the class (module)	Reference to directional effects <sup>1</sup>
EK_01	Describes the normal human karyotype and different types of sex determination.	C.W1.
EK_02	Knows the genetic causes of hereditary predisposition to cancer.	C.W2.
EK_03	Knows the principles of inheritance of a different number of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of extranuclear genetic information.	C.W3.
EK_04	Knows the genetic determinants of human blood groups and serological conflict in the Rh system.	C.W4.
EK_05	Knows the genetic determinants of the most common monogenic, polygenic and multifactorial diseases, basic chromosomal aberration syndromes, syndromes caused by	C.W5.

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<sup>1</sup>In the case of a path of education leading to obtaining teaching qualifications, also take into account the learning outcomes of the standards of education preparing for the teaching profession.

	genomic rearrangements, polymorphisms, epigenetic and post-transcriptional changes.	
EK_06	Knows the factors influencing the primary and secondary genetic balance of a population.	C.W6.
EK_07	Knows the genetic determinants of congenital malformations and selected rare diseases and the possibilities of their prevention.	C.W7.
EK_08	Is able to draw and analyze pedigrees and identify clinical-pedigree features suggesting a genetic basis of diseases.	C.U1.

### 3.3. Content curriculum

#### A. Problems of the lecture

Course contents
1. Basic concepts in genetics: genome structure, types of gene mutations, genetic polymorphisms, cell cycle regulation, mitosis, meiosis, apoptosis, telomeres
2. Monogenic heredity: autosomal dominant, autosomal recessive, X-linked - dominant, X-linked - recessive
3. Classical cytogenetics: human karyotype, numerical and structural chromosomal aberrations, selected syndromes of congenital defects, sex chromosome aneuploidies, methods of chromosome testing
4. Molecular cytogenetics: discussion of FISH, CGH and aCGH methods, MLPA, selected microdeletion syndromes
5. Molecular genetic methods and examples of their use: PCR-based methods, Sanger sequencing, next-generation sequencing
6. Genetics of cancer diseases: somatic mutations, proto-oncogenes, suppressor genes, DNA repair genes, theory Knudson, stages of carcinogenesis, carcinogens, personalized therapies in sporadic cancers
7. Hereditary cancer predisposition syndromes: characteristic features of hereditary syndromes, penetrance of mutations, expression of germline mutations, phenocopies, exemplary cancer predisposition syndromes (retinoblastoma, hereditary breast and ovarian cancer syndrome, hereditary nonpolyposis colon cancer, familial polyposis colonic, Li-Fraumeni syndrome)
8. Epigenetics: post-translational modifications of histones, DNA methylation, non-coding RNA, diseases resulting from epigenetic disorders, epigenetic therapies
9. Basic laws of inheritance – selected clinical examples. Population differentiation – “population genetics” and multifactorial inheritance

#### B. Problems of auditorium, seminar, laboratory and practical classes

Course contents
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Applications of classical chromosome testing methods. Indications for cytogenetic testing, chromosomal morphology, karyotype, karyotype, classical chromosomal test methods: GTG, CBG, RBG, Ag-NOR, HRT, principles of lymphocyte, fibroblast and amniocyte examination.
The importance of molecular cytogenetics in genetic testing. Fluorescent in-hybridization (FISH), types of probes, comparative genomic hybridization (CGH), microarray method, MLPA technique as a tool for the diagnosis of chromosomal aberrations.
Chromosomal aberrations that cause chromosomal syndromes. Division of chromosomal aberrations, causes of chromosomal aberration, mosaic and pseudomosaic character, record of chromosomal aberrations according to ISCN, effects of carrier of balanced and unbalanced aberrations, syndromes conditioned by chromosomal aberrations.
Molecular diagnosis of genetically conditioned diseases. Classification of genetic mutations, dynamic mutations, anticipation phenomena, mono-parental disomy - effects and effects, DNA methylation test, genetic causes of intellectual disability, genetic determinants of thrombophilia, genetic diagnosis of pregnancy failure, genetic determinants of Gilbert's syndrome, hemochromatosis, lactose intolerance, atopic dermatitis.
Recognition of types of inheritance of traits and human diseases - solving genetic crosses. Preparation and analysis of pedigrees.

### 3.4. Didactic methods

**Lecture:** problem lecture, lecture with multimedia presentation

**Exercises:** working in groups, solving tasks, discussion.

## 4. METHODS AND EVALUATION CRITERIA

### 4.1. Methods of verification of learning outcomes

Symbol of effect	Methods of assessment of learning outcomes (Eg.: tests, oral exams, written exams, project reports, observations during classes)	Form of classes
EK_01	observations during classes, colloquium	Lectures, Exercises
EK_02	observations during classes, colloquium	Lectures, Exercises
EK_03	observations during classes, colloquium	Lectures, Exercises
EK_04	colloquium, report	Exercises
EK_05	observations during classes, colloquium	Lectures, Exercises
EK_06	observations during classes, colloquium	Lectures, Exercises
EK_07	observations during classes, colloquium	Lectures, Exercises

EK_08	report	Exercises
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#### 4.2. Conditions for completing the course (evaluation criteria)

Positive evaluation of colloquium, positive assessment of reports, 100% attendance at classes.

Assessment criteria:

5.0 - has knowledge of the education content at the level of 93% -100%

4.5 - shows knowledge of the content of education at the level of 85% -92%

4.0 - shows knowledge of the content of education at the level of 77% -84%

3.5 - shows knowledge of the content of education at the level of 69% -76%

3.0 - shows knowledge of the content of education at the level of 60% -68%

2.0 - shows knowledge of the educational content below 60%

Positive evaluation of the subject can be obtained only on condition of obtaining a positive assessment for each of the established learning outcomes.

#### 5. TOTAL STUDENT WORKLOAD REQUIRED TO ACHIEVE THE DESIRED RESULT IN HOURS AND ECTS CREDITS

Activity	The average number of hours to complete the activity
Contact hours (with the teacher) resulting from the study schedule of classes	30
Contact hours (with the teacher) participation in the consultations, exams	3
Non-contact hours - student's own work (preparation for classes, exam, writing a paper, etc.)	27
SUM OF HOURS	60
TOTAL NUMBER OF ECTS	2

*\* It should be taken into account that 1 ECTS point corresponds to 25-30 hours of total student workload.*

#### 6. TRAINING PRACTICES IN THE SUBJECT

NUMBER OF HOURS	-
RULES AND FORMS OF APPRENTICESHIP	-

## 7. LITERATURE

### Basic literature:

1. Michael A. Lieberman, Rick Ricer. Biochemistry, Molecular Biology, and Genetics. Wolters Kluwer Health (JL). 2020.
2. Genetics Essentials: Concepts and Connections. Benjamin A. Pierce. Ed. 4. New York: W.H. Freeman and Company. 2018.

### Additional literature:

1. Cancer Genomics for the Clinician. Ramaswamy Govindan, Siddhartha Devarakonda. New York: Demos Medical Publishing. 2019.
2. From gene to therapy : understanding human disease through genetics. Michael Dean. [San Rafael, California]: Morgan & Claypool. 2017.
3. Color Atlas of Genetics. Eberhard Passarge. Georg Thieme (JL). 2018.
4. Medical genetics. Lynn B. Jorde, John C. Carey, Michael J. Bamshad. Philadelphia: Elsevier. 2020.

Acceptance Unit Manager or authorized person