

SYLLABUS

concerning the cycle of education 2022-2028

(date range)

Academic year 2022/2023

**1. BASIC INFORMATION CONCERNING THIS SUBJECT**

Subject	<b>General genetics</b>
Course code *	<b>Gen/C</b>
Faculty of (name of the leading direction)	<b>College of Medical Sciences</b>
Department Name	<b>Department of General and Clinical Genetics, Department of Biology</b>
Field of study	<b>medical direction</b>
level of education	<b>uniform master's studies</b>
Profile	<b>General academic</b>
Form of study	<b>stationary / extramural</b>
Year and semester	<b>year II, semester IV</b>
Type of course	<b>obligatory</b>
Language	<b>English</b>
Coordinator	<b>Prof. dr hab. n. med. Izabela Zawlik</b>
First and Last Name of the Teachers	<b>Prof. dr hab. n. med. Izabela Zawlik Dr n. med. Aleksander Myszka</b>

\* - 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	He knows the basic concepts in the field of genetics.	C.W1.
EK_02	Describes the phenomena of gene coupling and interactions.	C.W2.
EK_03	Describes the correct human karyotype and different types of gender determination.	C.W3.
EK_04	Describes the structure of chromosomes and the molecular basis of mutagenesis.	C.W4.
EK_05	He knows the principles of inheritance of various number of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of non-nuclear genetic information.	C.W5.
EK_06	He knows the genetic conditions of human blood groups and the serological conflict in the Rh system.	C.W6.

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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.

EK_07	Describes the aberrations of autosomes and heterosomes that cause disease, including cancers and oncogenesis.	C.W7.
EK_08	He knows the factors influencing the primary and secondary genetic balance of the population.	C.W8.
EK_09	He analyzes genetic crosswords and pedigrees of human traits and diseases, as well as evaluates the risk of a child's birth with chromosomal aberrations.	C.U1.

### 3.3 CONTENT CURRICULUM

#### A. Problems of the lecture

Course contents	Hours
Introduction - "man in the face of the puzzles of inheritance" - the development of basic science and its impact on worldview and human health; creation and development of genetics - great discoveries and the role of chance; participation of Poles.	2
The phenomenon of life - molecular ground - unity and diversity of forms; continuity of the species and evolutionary character of development depending on environmental conditions; the human genome, why we are similar - gene families.	2
Cell as an integral body of the organism; intercellular signaling; organization of genetic information in the cell; molecular basis of inheritance - division of somatic cells and germline and cell cycle - introduction to carcinogenesis.	3
Gen and its function and genetic variability - polymorphism and errors resulting from abnormalities in the material containing genetic information - chromosomal aberrations, gene mutations; epigenetic changes.	3
"Repairing errors" - "repair genes"; the possibilities and limitations of gene therapy.	3
Basic rights of inheritance - selected clinical examples.	2
Diversity of population - "population genetics" and multifactorial inheritance.	2
Basics of "genetic testing" - clinical examination and genetic tests.	3

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

Course contents	Hours
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.	2
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.	2

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.	2
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.	2
Recognition of types of inheritance of traits and human diseases - solving genetic crosses. Preparation and analysis of pedigrees.	2

### 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	colloquium	Lectures, Exercises
EK_02	colloquium	Lectures
EK_03	colloquium	Exercises
EK_04	colloquium	Lectures, Exercises
EK_05	colloquium	Lectures
EK_06	colloquium	Exercises
EK_07	colloquium	Lectures, Exercises
EK_08	colloquium	Lectures
EK_09	colloquium, report	Exercises

### 4.2 Conditions for completing the course (evaluation criteria)

Lectures (EK\_01, EK\_02, EK\_04, EK\_05, EK\_07, EK\_08)

Classes (EK\_01, EK\_03, EK\_04, EK\_06, EK\_07, EK\_09)

Positive evaluation of final colloquium and partial colloquiums, positive assessment of reports, 90% 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. BRS 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). 2017.

Acceptance Unit Manager or authorized person