

**SYLLABUS**  
**concerning the cycle of education 2023-2029**

(date range)

**1.1. BASIC INFORMATION CONCERNING THIS SUBJECT / MODULE**

Subject / Module	<b>General genetics</b>
Course code / module *	<b>Gen/C</b>
Faculty of (name of the leading direction)	<b>Medical College of Rzeszów University</b>
Department Name	<b>Chair of Molecular Medicine - Department of Genetics Department of Obstetrics - Department of Medical Genetics and Embryology;</b>
Field of study	<b>medical direction</b>
Level of education	<b>uniform master's studies</b>
Profile	<b>practical</b>
Form of study	<b>stationary / extramural</b>
Year and semester	<b>year II, semester IV</b>
Type of course	<b>obligatory</b>
Coordinator	<b>dr hab. prof. UR Izabela Zawlik</b>
First and Last Name of the Teacher	<b>dr hab. prof. UR Izabela Zawlik dr n. med. Antoni Pyrkosz dr n. med. Aleksander Myszka</b>

\* - According to the resolutions of the Faculty of Medicine

**1.2. Forms of classes, number of hours and ECTS**

Lecture	Exercise	Conversation	Laboratory	Seminar	ZP	Practical	Self-learning	Number of points ECTS
15	15	-	-	-	-	-	-	3

**1.3. The form of class activities**

☒ classes are in the traditional form

☐ classes are implemented using methods and techniques of distance learning

**1.4. Examination Forms / module** (exam, **credit with grade** or credit without grade)

**2. REQUIREMENTS**

The student should know the basics of biochemistry and molecular biology.
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### 3. OBJECTIVES, OUTCOMES, AND PROGRAM CONTENT USED IN TEACHING METHODS

#### 3.1. Objectives of this course/module

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 / MODULE (TO BE COMPLETED BY THE COORDINATOR)

EK (the effect of education)	The content of the learning effect defined for the subject (module)	Reference to directional effects (KEK)
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.
EK_07	Describes the aberrations of autosomes and heterosomes that cause disease, including ontogenesis cancers.	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.
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### 3.3 CONTENT CURRICULUM (filled by the coordinator)

#### A. Lectures

Course contents
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.
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.
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.
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.
"Repairing errors" - "repair genes"; the possibilities and limitations of gene therapy.
Basic rights of inheritance - selected clinical examples.
Diversity of population - "population genetics" and multifactorial inheritance.
Basics of "genetic testing" - clinical examination and genetic tests.

#### B. Exercises

Course contents
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-situ 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 TEACHING 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		Lectures, Exercises
EK_02		Lectures
EK_03		Exercises
EK_04		Lectures, Exercises
EK_05		Lectures
EK_06		Exercises
EK_07		Lectures, Exercises
EK_08		Lectures
EK_09		Exercises

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

<p>Lectures (EK_01, EK_02, EK_0_0, EK_05, EK_07, EK_08)</p> <p>Classes (EK_01, EK_02, EK_03, EK_02, EK_06, EK__07, EK_09)</p> <p>Positive evaluation of final colloquium and partial colloquiums, positive assessment of reports, 90% attendance at classes.</p>
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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	Hours / student work
Hours of classes according to plan with the teacher	30
Preparation for classes	20
Participation in the consultations	2
The time to write a paper / essay	-
Preparation for tests	25
Participation in colloquia	1
Other (e-learning)	-
SUM OF HOURS	78
TOTAL NUMBER OF ECTS	3

#### 6. TRAINING PRACTICES IN THE SUBJECT / MODUL

Number of hours	-
Rules and forms of apprenticeship	-

#### 6. LITERATURE

##### READING:

1. Genetyka Medyczna – Tobias ES, Connor M, Ferguson-Smith M. przekład pod red.A. Latos-Bieleńskiej PZWL, 2013.
2. Jorde L.B., Carey J.C., Bamshad M.J., White R.L.: Genetyka medyczna. Wyd. II, red. wyd. pol. Bogdan Kałużewski, 2013
3. Badania cytogenetyczne w praktyce klinicznej, A. Tomaszewska, M. Srebniak Wydawnictwo Lekarskie PZWL, 2008

Additional literature:

1. Biologia molekularna w medycynie. Elementy genetyki klinicznej pod redakcją J. Bala. PWN Warszawa 2008 i nowsze wydania
2. Genetyka medyczna. Notatki z wykładów. John R. Bradley, David R. Johnson, Barbara R. Pober, red. wyd. pol. Tadeusz Mazurczak, PZWL, 2008
3. Genetyka. JM Friedman pod red. J. Limona. U&P 1997.

Acceptance Unit Manager or authorized person