

SYLLABUS

Concerning the cycle of education **2025- 2031**

Academic year 2026/2027

1. BASIC INFORMATION CONCERNING THIS SUBJECT

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|---------------------------------------|---------------------------------------------|
| Subject / Module | General genetics |
| Course code* | Gen/C |
| Faculty (name of the leading faculty) | Faculty of Medicine, University of Rzeszow |
| Department Name | Department of General and Clinical Genetics |
| Field of study | Medicine |
| Level of education | Uniform master studies |
| Profile | General academic |
| Form of study | Stationary / non-stationary |
| Year and semester | Year II, semester 3 |
| Type of course | Obligatory |
| Language | English |
| Coordinator | Prof. Izabela Zawlik, PhD |
| Name(s) of the instructor(s) | Dr Aleksander Myszkowski, PhD |

* - according to the resolutions of Educational Unit

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

| Semester No. | Lecture | Exercise | Conversation | Laboratory | Seminar | Practical | Other | Number of points ECTS |
|--------------|---------|----------|--------------|------------|---------|-----------|-------|-----------------------|
| 3 | 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); credit with grade

2. BASIC REQUIREMENTS

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| The student should know the basics of biochemistry. |
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3. OBJECTIVES, LEARNING OUTCOMES, AND PROGRAM CONTENT USED IN TEACHING METHODS

3.1. Objectives of this course

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| 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 ¹ |
|------------------------------|------------------------------------------------------------------------------------|-----------------------------------------------|
| 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. |

¹In 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.

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| 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 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

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| 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, effects of carrier of balanced and unbalanced aberrations, selected syndromes of congenital defects, sex chromosome aneuploidies, record of chromosomal aberrations according to ISCN, 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) |

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| 8. Epigenetics: post-translational modifications of histones, DNA methylation, non-coding RNA, diseases resulting from epigenetic disorders, epigenetic therapies |
| SKILLS: THE GRADUATE IS ABLE TO |
| 9. Basic laws of inheritance – selected clinical examples. Population differentiation – “population genetics” and multifactorial inheritance |

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

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| 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. 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, colloquium, 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, report | Exercises |
| EK_09 | colloquium, report | Exercises |

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 INTENDED LEARNING OUTCOMES IN HOURS AND ECTS CREDITS

| Form of activity | Average number of hours required to complete the activity |
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|------------------|-----------------------------------------------------------|

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| 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

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| Number of hours | - |
| Rules and forms of internships | - |

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.

Approved by the Head of the Department or an authorised person