Appendix number 1.5 to The Rector UR Resolution No. 12/2019

SYLLABUS

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

(date range)

* 1. BASIC INFORMATION CONCERNING THIS SUBJECT / MODULE

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| --- | --- |
| Subject / Module | Clinical genetics |
| Course code / module \* | GenK/C |
| Faculty of (name of the leading direction) | Medical College of Rzeszów University |
| Department Name | Department of Molecular Medicine, Department of Genetics;  Department of Obstetrics - Department of Medical Genetics and Embryology; |
| Field of study | medical direction |
| Level of education | uniform master's studies |
| Profile | practical |
| Form of study | stationary / extramural |
| Year and semester | year III, semester V |
| Type of course | obligatory |
| Coordinator | lek. med. Paweł Zapolnik |
| First and Last Name of the Teacher | lek. med. Paweł Zapolnik |

\* - 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 | - | - | - | - | - | - | 2 |

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

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| Anatomy, biochemistry, physiology and pathophysiology |

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

* 1. Objectives of this course/module

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| --- | --- |
| C1 | Transfer of knowledge on the basics of clinical genetics and molecular biology techniques and their application |
| C2 | Diagnosis of genetic diseases. The importance of consultation and genetic research  in the most common diseases genetically determined |

3.2 OUTCOMES FOR THE COURSE / MODULE (TO BE COMPLETED BY THE COORDINATOR)

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| --- | --- | --- |
| EK (the effect of education) | The content of the learning effect defined for the subject (module) | Reference to directional effects (KEK) |
| EK­\_01 | describes the aberrations of autosomes and heterosomes that cause disease, including tumors of oncogenesis | C.W7 |
| EK\_02 | knows the basics of diagnosis of gene and chromosomal mutations responsible for hereditary and acquired diseases, including cancer | C.W9 |
| EK\_03 | determines benefits and threats resulting from the presence of genetically modified organisms (GMOs) in the ecosystem | C.W10 |
| EK\_04 | knows the genetic mechanisms of acquiring drug resistance by microbes and cancer cells | C.W11 |
| EK\_05 | knows and understands the causes, symptoms, principles of diagnosis and therapeutic treatment in the most common inherited diseases | E.W35 |
| EK\_06 | knows the basic directions of therapy development, in particular the possibilities of cell therapy, gene therapy and targeted therapy in specific diseases | C.W41 |
| EK\_07 | identifies indications for prenatal testing | C.U2 |
| EK\_08 | can decide on the need for cytogenetic and molecular tests | C.U3 |
| EK\_09 | performs morphometric measurements, analyzes the morphogram and records karyotypes of diseases | C.U4 |
| EK\_10 | estimates the risk of a given disease becoming apparent in the offspring based on family predisposition and the influence of environmental factors | C.U5 |

**3.3 CONTENT CURRICULUM (filled by the coordinator)**

1. **Lectures**

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| **Course contents** |
| 1. A reminder of the basics of genetic / hereditary information and methods of inheritance - inheritance by classical laws and atypical forms of inheritance |
| 2. Clinical basis of genetic diagnosis - genetic interview, morphological phenotype - dysmorphological assessment, imaging diagnostics, biochemical, cytogenetic, cytogenetic and molecular, genetic tests |
| 3. Genetics of development and congenital malformations - their conditioning |
| 4. Personalized medicine - the basics of pharmacogenetics and strategies of medical therapies |
| 5. Immunogenetics and oncogenetics |
| 6. Delay in development, cognitive disorders, emotions and expression of emotions, motivation and thinking; neurogenetics - selected issues |
| 7. Orphan diseases - based on congenital metabolism defects |
| 8. Clinical genetics in various medical specialties and the basics of genetic counseling |

**B**. **Exercises**

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| Course contents |
| 1. Introduction - elements of subjective examination - "genetic interview". |
| 2. Introduction - the basics of physical-subject examination. Proper individual development. Normal and incorrect features - introduction to dysmorphological diagnosis. |
| 3. Professional online databases - Online Mendelian Inheritance in Man [OMIM]; The Online Metabolic and Molecular Bases of Inherited Disease [OMMBIB]; Orohanet; NEUROMUSCULAR DISEASE CENTER; Retinal Information Network [RetNet]; other) and dedicated - specialized databases for dysmorphological and neurogenetic assessment (LMD - London Medical Databases: LDDB - London Dysmorphology Database, London Dysmorphology Database, London Neurogenetics Database & Dysmorphology Photo Library on CD-ROM, GENEEYE - The London Ophthalmic Genetics Database ; POSSUM - Pictures of Standard Syndromes and Undiagnosed Malformations, SYNDROC); FACE2GENE and others; "Genetic atlases". How to use this data in practice. |
| 4. Characteristic clinical features occurring in people with unsustainable chromosomal aberrations - concerning quantitative and structural disorders of chromosomes, recognized using classical cytogenetics. |
| 5. Characteristic clinical features occurring in people with adjacent gene disorders. Characteristic clinical features in selected cases of epigenetic disorders. |
| 6. Basic clinical features and diagnostics in selected monogenic disorders - with autosomal dominant character. |
| 7. Basic clinical features and diagnostics in selected monogenic disorders - with autosomal recessive character. |
| 8. Basic clinical features and diagnostics in selected monogenic disorders coupled with gonosomes. |
| 9. Clinical genetics in "Gynecology and Obstetrics", "Andrology", "Sexology". Genetic aspects of pregnancy failure and primary infertility - reproductive problems - male and female infertility, recurrent miscarriages and stillbirths; disorders of sex development (disorders of sex development - DSD); sex and gender, transsexuality - transgenderism, homosexuality. |
| 10. Clinical genetics in "Perinatology", "Neonatalogy", "Pediatrics". Developmental biology - congenital malformations - etiology, diagnostic procedures on selected examples. |
| 11. Clinical genetics in "Metabolic Pediatrics", "Clinical Biochemistry" and "Endocrinology". Selected metabolic and endocrine disorders - basic clinical conditions, with special emphasis on "emergencies". |
| 12. Clinical genetics in "Neurology", "Psychiatry". Selected neurological conditions and mental disorders - their conditioning, diagnostics. |
| 13. Clinical genetics in "Oncology". "Hereditary neoplasms" - proceedings - prophylaxis, diagnostics, targeted therapy. |
| 14. Clinical Genetics in various disciplines / medical specialties, in the "Practice of the Family Doctor". Genetic predisposition and the influence of environmental factors, including lifestyle, in adult diseases, on the example of the most common "comprehensive" - ​​"civilization" diseases. |
| 15. Genetic counseling and prenatal diagnosis - selected examples, indications for prenatal diagnosis. Ethical dilemmas related to clinical genetics. |

**3.4 TEACHING METHODS**

**Lectures -** problem lecture / lecture with multimedia presentation

**Exercises**- working in groups / solving tasks / discussion / discussing clinical cases

**Student's own work:** work with a book

4 METHODS AND EVALUATION CRITERIA

4.1 Methods of verification of learning outcomes

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| 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 | written exam, colloquium | Lectures, Exercises |
| Ek\_ 02 | written exam, colloquium | Lectures, Exercises |
| EK­\_03 | written exam, colloquium | Lectures, Exercises |
| EK­\_04 | written exam, colloquium | Lectures, Exercises |
| EK­\_05 | written exam, colloquium | Lectures, Exercises |
| EK­\_06 | written exam | Lectures |
| EK­\_07 | test | Exercises |
| EK­\_08 | written exam, colloquium | Lectures, Exercises |
| EK­\_09 | written exam, colloquium | Lectures, Exercises |
| EK­\_10 | written exam, colloquium | Lectures, Exercises |

4.2 Conditions for completing the course (evaluation criteria)

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| Positive evaluation of the exam and tests, 90% attendance.  Positive evaluation of the subject can be obtained only on condition of obtaining a positive assessment for each of the established learning outcomes.  **Lectures**  The condition to take the exam is to get a positive grade from the exercises.  Evaluation criteria:  **Knowledge assessment (EK\_01-EK\_05):**  5.0 - has knowledge of each of the contents of education at the level of 90% -100%  4.5 - has knowledge of each of the content of education at the level of 84% -89%  4.0 - has knowledge of each of the content of education at the level of 77% -83%  3.5 - has knowledge of each of the content of education at the level of 70% -76%  3.0 - has knowledge of each of the content of education at the level of 60% -69%  2.0 - has knowledge of each of the contents of education below 60%  **Exercises**:  - written exam  - observation of the student's work  - assessment of student activity during classes  - discussion during classes  **Skill assessment (EK\_06-EK\_10):**  5.0 - the student actively participates in classes, is well prepared, knows the basic concepts of genetics very well, analyzes genetic crosses, correctly identifies indications for prenatal tests  4.5 - the student actively participates in classes,  knows the basic concepts of genetics to a good degree, analyzes genetic crosses, correctly identifies indications for prenatal tests  4.0 - the student actively participates in classes, is improved, knows the basic concepts of genetics, analyzes genetic crosses, identifies indications for prenatal tests, sometimes making mistakes  3.5 - the student participates in the classes, his scope of preparation does not allow for a comprehensive presentation of the discussed problem, he knows the basic concepts of genetics to a sufficient degree, analyzes genetic crosswords, identifies indications for prenatal tests by making mistakes  3.0 - the student participates in classes, sufficiently knows the basic concepts of genetics, analyzes genetic crosswords, identifies indications for prenatal tests often making mistakes  2.0 - the student passively participates in the classes, has not sufficiently mastered the basic concepts of genetics, analyzes genetic crosswords often making mistakes, incorrectly identifies indications for prenatal tests |

5. Total student workload required to achieve the desired result in hours and ECTS credits

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| --- | --- |
| Activity | Hours / student work |
| Hours of classes according to plan with the teacher | 30 |
| Preparation for classes | 15 |
| Participation in the consultations | 2 |
| The time to write a paper / essay | - |
| Preparation for tests | 10 |
| Participation in colloquia | 1 |
| Other (e-learning) | - |
| SUM OF HOURS | 58 |
| TOTAL NUMBER OF ECTS | **2** |

6. TRAINING PRACTICES IN THE SUBJECT / MODUL

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

1. LITERATURE

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| **READING:**  1. Genetyka Medyczna – Tobias ES, Connor M., Ferguson-Smith M. przekład pod red. A. Latos-Bieleńskiej PZWL Warszawa 2013.  2. Genetyka medyczna - Jorde L.B., Carey J.C., Bamshad M.J., White R.L.: Wyd. II, red. wyd. pol. Bogdan Kałużewski, 2013  3. Genetyka człowieka. Rozwiązywanie problemów medycznych. Bruce R. Korf. PWN Warszawa 2003. Przekład pod redakcją A Pawlaka. |
| Additional literature:   1. Dziecko z zespołem wad wrodzonych. Diagnostyka dysmorfologiczna. L. Korniszewski. PZWL 2005. 2. Onkogenetyka - teoria i praktyka kliniczna. A. Stembalska, K. Pesz, M.M. Sąsiadek, UM Wrocław Wydawnictwo Wrocław 2015 3. Vademecum Metabolicum Podręcznik pediatrii metabolicznej – Zschocke / Hoffmann, wyd. II w tłumaczeniu Ewy Pronickiej; wydawniectwo „milupa”, projekt PERFECT Warszawa 2005   4. Kompendium Pediatrii Praktycznej pod red. A. Jankowskiego. Cornetis 2010 |

Acceptance Unit Manager or authorized person