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

SYLLABUS

concerning the cycle of education 2022-2028

(date range)

1.1. BASIC INFORMATION CONCERNING THIS SUBJECT / MODULE

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	**dr n. med. Antoni Pyrkosz**
First and Last Name of the Teacher	**dr n. med. Antoni Pyrkosz**

* - 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

Anatomy, biochemistry, physiology and pathophysiology

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

3.1. Objectives of this course/module

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)

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)

A. Lectures

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

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

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)

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

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

Number of hours	-
Rules and forms of apprenticeship	-

6. LITERATURE

READING:
1. Tobias ES, Connor M, Ferguson-Smith M. Essential Medical Genetics, 6th Edition. Wiley-Blackwell, Hoboken, NJ, USA; 2011.
2. Jorde LB, Carey JC, Bamshad MJ. Medical Genetics. Elsevier Health Sciences, Amsterdam, The Netherlands; 2019.
3. Jones KL, Jones MC, Del Campo M. Smith's Recognizable Patterns of Human Malformation, 8th Edition. Elsevier, Amsterdam, The Netherlands; 2021.
4. Saul RA. Medical Genetics in Pediatric Practice. American Academy of Pediatrics; 2013.
5. Bradley Schaefer G., Thompson JN Jr. Medical Genetics: An Integrated Approach. McGraw-Hill Education Ltd; 2014.
6. Turnpenny P, Ellard S, Cleaver R. Emery's Elements of Medical Genetics and Genomics. Elsevier; 2021.
7. Read A, Donnai D. New Clinical Genetics, fourth edition. A guide to genomic medicine. Scion Publishing Ltd; 2020.
8. Nussbaum R, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. Elsevier; 2015.
9. Pritchard DJ, Korf BR. Medical Genetics at a Glance. Wiley-Blackwell; 2013.
10. Chandar N, Viselli S. Lippincott Illustrated Reviews: Cell and Molecular Biology. Wolters Kluwer Health; 2023.
11. Adkison LR. Elsevier's Integrated Review Genetics. Elsevier; 2012.
12. Goldberg S. Clinical Genetics Made Ridiculously Simple. MedMaster Corporation; 2020.
13. Passarge E. Color Atlas of Genetics. Georg Thieme; 2018.
14. Zschocke J, Hoffmann GF. Vademecum Metabolicum. Diagnosis and Treatment of Inborn Errors of Metabolism. Georg Thieme; 2021.

Additional literature:
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.
4. Dziecko z zespołem wad wrodzonych. Diagnostyka dysmorfologiczna. L. Korniszewski. PZWL 2005.
5. Onkogenetyka - teoria i praktyka kliniczna. A. Stembalska, K. Pesz, M.M. Sąsiadek, UM Wrocław Wydawnictwo Wrocław 2015
6. Vademecum Metabolicum Podręcznik pediatrii metabolicznej – Zschocke / Hoffmann, wyd. II w tłumaczeniu Ewy Pronickiej; wydawniectwo „milupa", projekt PERFECT Warszawa 2005
7. Kompendium Pediatrii Praktycznej pod red. A. Jankowskiego. Cornetis 2010

Acceptance Unit Manager or authorized person