

SYLLABUS

Concerning the cycle of education 2018 - 2024

Academic year 2020/2021

1. BASIC INFORMATION CONCERNING THIS SUBJECT

Subject	Clinical Genetics
Course code *	CGen
Faculty of (name of the leading direction)	Faculty of Medicine
Department Name	Collegium of Medical Science, Rzeszow University / Department of Genetics
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 VI
Type of course	obligatory
Language	English
Coordinator	MD PhD Antoni Pyrkosz
First and Last Name of the Teachers	MD PhD Antoni Pyrkosz, M.Sc. Magdalena Dziorek

* - according to the resolutions of Educational Unit

1.1. Forms of classes, number of hours and ECTS

Semester No.	Lecture	Exercise	Conversation	Laboratory	Seminar	ZP	Practical	Other	Number of points ECTS
VI	15	15	-	-	-	-	-	-	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 anatomy, physiology, pathophysiology, biochemistry and molecular biology.

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

3.1 OBJECTIVES OF THIS COURSE

To impart knowledge of the fundamentals of clinical genetics and molecular biology techniques and their application.
Diagnostics of genetic diseases. The importance of consultation and genetic testing in the most common genetically determined disorders.

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	Knowledge of the basis for diagnosis of gene and chromosome mutations responsible for inherited and acquired diseases, including cancer. Description of the phenomena of gene coupling and interactions.	C.W1.
EK_02	Identify the need for prenatal tests. Able to decide on the need to perform cytogenetic and molecular tests.	C.W2.
EK_03	Know and understand the causes, symptoms, principles of diagnosis and therapeutic management of the most common hereditary diseases.	C.W3.
EK_04	Describe the benefits and risks associated with the presence of genetically modified organisms (GMOs) in the ecosystem. Know the genetic mechanisms for the acquisition of drug resistance by microorganisms and cancer cells.	C.W4.
EK_05	Describe the aberrations of autosomes and heterosomes that cause disease, including ontogenesis cancers. Perform morphometric measurements, analyse the morphogram and record karyotypes of diseases.	C.W5.
EK_06	Know the basic directions of therapy development, in particular the possibilities of cell, gene and targeted therapy in specific diseases. Analysis of genetic crosswords and pedigrees of human traits and diseases, and risk associate with chromosomal aberrations from birth.	C.W6.
EK_07	Estimate the risk of the appearance of a given disease in offspring on the basis on family predisposition and the influence of environmental factors. Wellbeing and putting patients first. Designing	C.W7. C.U1.

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.

	and practicing patient centred care.	
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3.3 CONTENT CURRICULUM

A. Lecture topics.

Course contents
1. Introduction to genetics – hereditary information and inheritance. Understanding human genetic variation. Unique DNA.
2. The clinical basis of genetic diagnostics - genetic history; morphological phenotype - dysmorphological evaluation, imaging, biochemical, cytogenetic, cytogenetic-molecular diagnostics, genetic tests.
3. Developmental genetics and congenital malformations - their determinants.
4. The vision of personalised medicine - basics of pharmacogenetics and strategies of medical therapies. Immunogenetics and oncogenetics.
5. Developmental delay, cognitive disorders, emotions and expression of emotions, motivation or thinking; neurogenetics - selected issues.
6. Orphan diseases - based on inborn defects of metabolism.
7. Clinical genetics in various medical specialties and the basis of genetic counselling.

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

Course contents
1. Introduction - basics of physical-objective examination. History of genetics. Individual development. Normal versus abnormal features - introduction to dysmorphological diagnosis. Professional online databases. How to use these data and applications in the daily routine of medical practice?
2. Clinical characteristics of individuals with unbalanced chromosome aberrations - concerning quantitative and structural chromosome abnormalities, recognised using classical cytogenetic techniques. Characteristic clinical features in subjects with “adjacent gene” abnormalities. Characteristic clinical features in selected cases of epigenetic disorders.
3. Basic clinical features and diagnostics in selected monogenic disorders – autosomal dominant and recessive character. Monogenic disorders are coupled with genosomes.
4. Clinical genetics in Gynaecology 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 (DSD); transsexualism - transgenderism, homosexuality.
5. Clinical genetics in Perinatology, Neonatology, Pediatrics. Developmental biology congenital malformations – etiology, diagnostic management on selected examples. Clinical genetics in Metabolic Pediatrics, Clinical Biochemistry and Endocrinology. Selected metabolic and endocrine disorders – basic clinical conditions, with special attention to “emergency conditions”.

6. Clinical genetics in Neurology and Psychiatry. Selected neurological states and mental disorders - their conditions, diagnosis. Clinical genetics in Oncology. Hereditary tumours – management, prevention, diagnosis and targeted therapy.
7. Clinical Genetics in various medical disciplines/specialties, in “Family Physician Practice”. Genetic predispositions and the influence of environmental factors, including lifestyle, in adult diseases, on the example of the most frequent complex – civilization diseases. Genetic counselling and prenatal diagnostics – selected examples, indications for prenatal diagnostics. Ethical dilemmas related to clinical genetics.

3.4 Didactic methods

Lecture: problem lecture, lecture with multimedia presentation

Exercises: working in groups, solving tasks, discussion, preparation of presentation.

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

4.2 Conditions for completing the course (evaluation criteria)

<p>Lectures (EK_01, EK_02, EK_03, EK_04, EK_05, EK_06, EK_07) Classes (EK_01, EK_02, EK_03, EK_04, EK_05, EK_06, EK_07)</p> <p>Positive evaluation of final colloquium and partial colloquiums, positive assessment of reports, 90% attendance at classes.</p> <p>Assessment criteria:</p> <p>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%</p> <p>Positive evaluation of the subject can be obtained only on condition of obtaining a positive assessment for each of the established learning outcomes.</p>

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.)	45
SUM OF HOURS	78
TOTAL NUMBER OF ECTS	3

** 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. LYNN B. JORDE, JOHN C. CAREY, MICHAEL J. BAMSHAD, MEDICAL GENETICS, PHILADELPHIA, 2020.
2. Michael A. Lieberman, Rick Ricer. BRS Biochemistry, Molecular Biology, and Genetics. Ed. 7. Wolters Kluwer Health (JL), 2019.
3. Benjamin Pierce. Genetics Essentials: Concepts and Connections. Ed. 4. Macmillan International Higher Education (JL), 2018.
4. Benjamin Pierce. Genetics: A Conceptual Approach. Ed. 6. Macmillan International Higher Education (JL), 2016.

Additional literature:

1. Cancer Genomics for the Clinician. Ramaswamy Govindan, Siddhartha Devarakonda. Eurospan (JL), 2019.
2. Color Atlas of Genetics. Eberhard Passarge. Georg Thieme (JL). 2017.

Acceptance Unit Manager or authorized person.