UNIVERSITY OF WARMIA AND MAZURY IN OLSZTYN

of Medicine

Course/module syllabus - part A

48SJ-CLINGEN
ECTS: 2
YEAR: 2023L

CLINICAL GENETICS
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Course/module:
Clinical Genetics
Fields of education:

Course status:	mandatory
Course group:	B - przedmioty kierunkowe
ECTS code:	
Field of study:	Medicine
Specialty area:	Medicine
Educational profile:	General academic
Form of study:	full-time
Level of study:	uniform master's studies
Year/semester:	4 / 8

Type of course:
Classes, Seminar, Lecture

Number of hours per semester/week:	Classes: 10, Seminar: 15, Lecture: 15

Teaching forms and methods
Classes(K1, U1, W1) : Auditorium exercises - cases of genetically determined diseases, development of genetic counseling, clinical methodology, Seminar(K1, U1, W1) : Presentation with discussion of self-developed topics by students, Lecture(K1, U1, W1) : Theoretical lectures using audiovisual presentations and problem methods (case method, didactic discussion, brainstorming, decision tree)

Form and terms of the verification results:
CLASSES: Prezentation - null(K1, U1, W1) ; SEMINAR: Colloquium test - null(K1, U1, W1) ; LECTURE: Colloquium test - null(K1, U1, W1)

Number of ECTS points:	2
Language of instruction:	English

Introductory courses:
genetics and molecular biology, physiology, pathophysiology, embryology
Preliminary requirements:
completing the introductory courses

Name of the organizational unit offering the course:
Katedra Fizjologii i Patofizjologii Człowieka,
Person in charge of the course:
dr n. med. Małgorzata Pawłowicz,
Course coordinators:

Notes:
It is possible to systematically expand the knowledge acquired during compulsory classes by participating in meetings and scientific works of the Student Science Club

COURSE CONTENT
CLASSES

The thematic scope of exercises and seminars remains related to the topic of lectures. Seminars and exercises are aimed at consolidating the thematic scope of lectures and learning the practical use of knowledge acquired during the lecture block. Thematic scope of exercises: Pedigree analysis. Morphological phenotype analysis. Cytogenetics. Selected syndromes genetically determined in clinical practice. Thematic scope of the seminars: Genetic counseling. Chromosome aberrations. Microdeletion syndromes. Sex differentiation disorders. Phacomatoses, neurogenetics. Genetic prenatal diagnosis. Oncogenetics.

LECTURES

1. Basic concepts and definitions: DNA structure, DNA replication, transcription and translation, chromosome structure, cell cycle and cell division. Types of mutations and their consequences. Basic molecular diagnostic tests. Anomalies in the number and structure of chromosomes. Basic methods of cytogenetic diagnostics. Types of dysmorphic traits, mechanism and etiology of developmental malformations. Rules for conducting family interviews and pedigree preparation. 2. Diseases caused by chromosomal abnormalities: autosomal aneuploidy, sex chromosome aneuploidy, polyploidies, deletion and microdeletion syndromes, syndromes associated with chromosome instability. The concept of epigenetics and genomic imprinting. 3. One-gen inheritance (Mendelian): autosomal dominant, autosomal recessive, dominant and recessive sex-linked, atypical - discussion on examples of selected diseases. 4. Mitochondrial diseases conditioned by mitochondrial and nuclear DNA mutations. Metabolic diseases. Neurogenetics. 5. Multigenic inheritance, relationships between genetic predisposition and environmental factors - multifactorial diseases. The development of genetics and new disease classifications and therapeutic options - personalized medicine: discussion on clinical cases. Summary of the block of lectures on clinical genetics.

EDUCATIONAL OBJECTIVE:

Understanding the principles of inheritance, etiology, symptomatology and medical management in genetic diseases. Mastering specialized dysmorphological denomination and rules for describing and interpreting results of genetic tests. To learn and put into practice the skill of determining indications for performing genetic tests in pre- and postnatal diagnostics. Mastering the skills of anamnesis, physical examination and genetic counseling. Acquaintance with ethical, moral and legal problems related to genetic diagnostics and counseling. The ability to recognize a genetic disease and develop genetic counseling. Obtaining modern and up-to-date knowledge in the field of classic and novel methods of cytogenetic and molecular diagnostics (karyotype, FISH, MLPA, microarrays, Sanger sequencing, next generation sequencing with particular emphasis on whole exome sequencing). Getting to know the possibilities of primary, secondary and tertiary prevention of selected hereditary diseases.

DESCRIPTION OF LEARNING OUTCOMES FOR THE COURSE IN RELATION TO FIELD AND MAJOR LEARNING OUTCOMES

Codes of learning outcomes in a major field of study:	M/NM+++,
Codes of learning outcomes in a major area of study:	B.U13.+, B.W.28.+, B.W13.+, B.W14.+, C.U1.+, C.U2.+, C.U3.+, C.U4.+, C.U5.+, C.W1.+, C.W2.+, C.W3.+, C.W4.+, C.W5.+, C.W6.+, C.W7.+, C.W8.+, C.W9.+, D.U17.+, D.U6.+, D.U7.+, E.W1.+, E.W37.+, K.1.+, K.2.+, K.3.+, K.6.+,

LEARNING OUTCOMES:

Knowledge

W1 - Basic definitions in genetics. Knowledge of the structure of DNA, RNA and chromatin. Knowledge of intercellular interactions, basic cell signaling pathways and their disorders in specific diseases. Knowledge of basic processes at the cellular level. Understanding the function of the genome, transcriptome and proteome and laboratory methods for their study. Ability to search interactions between genes. Basic knowledge about Mendelian and non-Mendelian inheritance. Knowledge of bioinformatics methods used in the modern genetic analyzes. The ability to describe the correct human karyotype, select appropriate diagnostic methods to recognize chromosomal aberrations, mono- / oligo- / poligenetic disorders or diseases associated with DNA methylation disorders. Understanding the basics of population genetics. Knowledge of the genetic conditions of human blood groups.

Skills

U1 - The ability to choose cytogenetic and molecular analysis depending on the patient's phenotype. Ability to inform the patient about the purpose, course and possible risk of the proposed genetic tests and to obtain the patient's informed consent. Ability to create family tree, interpret them and estimate the risk of disease recurrence in the patient's family. The ability to identify indications for prenatal examinations. Ability to analyze karyotypes and molecular research results. The ability to provide the patient and his family with unfavorable information on the prognosis and risk of disease recurrence. The ability to use modern free available genetic databases, search and critical literature analysis in terms of credibility and quality of scientific data.

Social competence

K1 - Knowledge of patient rights and medical confidentiality. The ability to manage and prioritizing the patient's well-being. The ability to establish and maintain full respect of contact with a patient with a genetically determined disease, with particular emphasis on neurodegenerative diseases. Ability to follow the rules of medical and

academic ethics.

BASIC LITERATURE

1) Lynn B. Jorde,‎ John C. Carey MD MPH,‎ Michael J. Bamshad MD, Genetyka medyczna., wyd. Elsevier, 2015 ; 2) Connor JM, Ferguson-Smith MA, Podstawy genetyki medycznej., wyd. Wydawnictwo Lekarskie PZWL, 1998 ; 3) Friedman JM i wsp., Genetyka., wyd. Urban & Partner, 2000 ; 4) Korniszewski L., Dziecko z zespołem wad wrodzonych. Diagnostyka dysmorfologiczna., wyd. Wydawnictwo Lekarskie PZWL, 2005 ; 5) Kapelańska-Pręgowska J., Prawne i bioetyczne aspekty testów genetycznych., wyd. Wolters Kluwer Polska, 2011

SUPPLEMENTARY LITERATURE

1) Bennett RL., The Practical Guide to the Genetic Family History., wyd. Wiley Blackwell, 2010 ; 2) Darras BT, Jones HR, Ryan MM, De Vivo DC., Neuromuscular Disorders of Infancy, Childhood, and Adolescence, Second Edition: A Clinician's Approach., wyd. Academic Press, 2014 ; 3) Demkow U, Ploski R., Applications for Next-Generation Sequencing., wyd. Academic Press, 2016 ; 4) Firth HV, Hurst JA., Oxford Desk Reference Clinical Genetics., wyd. Oxford University Press, 2017 ; 5) Jones KL, Jones MC, Del Campo M., Smith's Recognizable Patterns of Human Malformation., wyd. Elsevier, 2013 ; 6) Pevsner J., Bioinformatics and Functional Genomics., wyd. Wiley Blackwell, 2015 ; 7) Rosenberg RN, Pascual JM., Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease., wyd. Academic Press, 2015 ; 8) Wood N., Neurogenetics: A Guide for Clinicians., wyd. Cambridge University Press, 2012 ; 9) Trygve T., Handbook of Epigenetics: The New Molecular and Medical Genetics., wyd. Academic Press, 2011 ; 10) Sadler TW., Langman's Medical Embryology., wyd. LWW, 2015

Detailed description of the awarded ECTS points - part B

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The awarded number of ECTS points is composed of:

1. Contact hours with the academic teacher:

- participation in: classes	10 h.
- participation in: seminar	15 h.
- participation in: lecture	15 h.
- consultation	2 h.
	42 h.

2. Student's independent work:

- analysis of current literature on selected disease entities for later active participation in discussion during classes.	8 h.
	8 h.

1 ECTS point = 25-30 h of the average student's work, number of ECTS points = 50 h : 25 h/ECTS = 2,00 ECTS on average: **2 ECTS**

- including the number of ECTS points for contact hours with direct participation of the academic teacher: 1,68 ECTS points,
- including the number of ECTS points for hours completed in the form of the student's independent work: 0,32 ECTS points,