UNIVERSITY OF WARMIA AND MAZURY IN OLSZTYN
Faculty of Medicine

Course sylabus – part A
Clinical Genetics

48SJ-GENKL
ECTS: 2.00
CYCLE: 2023L

SUBJECT MATTER CONTENT

LECTURE
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.

SEMINAR
The thematic scope of seminars remains related to the topic of lectures. Seminars are aimed at consolidating the thematic scope of lectures and learning the practical use of knowledge acquired during the lecture block. Thematic scope of the seminars: Genetic counseling. Chromosome aberrations. Microdeletion syndromes. Sex differentiation disorders. Phacomatoses, neurogenetics. Genetic prenatal diagnosis. Oncogenetics.

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.

TEACHING 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

Legal acts specifying learning outcomes: 3112022, 672/2020
Disciplines: medical sciences
Status of the course: Obligatoryjny
Group of courses: A - przedmioty podstawowe
Code: ISCED 0912
Field of study: Medicine, Medicine
Scope of education:
Profile of education: General academic
Form of studies: full-time
Level of studies: uniform master's studies
Year/semester: 4/8

Types of classes: Lecture, Seminar, Classes
Number of hours in semester: Lecture: 15.00, Seminar: 15.00, Classes: 10.00
Language of instruction: Polish
Introductory subject: genetics and molecular biology, physiology, pathophysiology, embryology
Prerequisites: completing the introductory courses

Name of the organisational unit conducting the course: Katedra Pediatrii Klinicznej
Person responsible for the realization of the course: dr n. med. Małgorzata Pawłowicz
e-mail: malgorzata.pawlowicz@uwm.edu.pl

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

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 THE LEARNING OUTCOMES OF THE COURSE IN RELATION TO THE DESCRIPTION OF THE CHARACTERISTICS OF THE SECOND LEVEL LEARNING OUTCOMES FOR QUALIFICATIONS AT LEVELS 6-8 OF THE POLISH QUALIFICATION FRAMEWORK IN RELATION TO THE SCIENTIFIC DISCIPLINES AND THE EFFECTS FOR FIELDS OF STUDY:

Symbols for outcomes related to the discipline:	M/NM+++, M/NMA_P7S_KR+++, M/NMA_P7S_KO+++, M/NMA_P7S_WG+++, M/NMA_P7S_UW+++
Symbols for outcomes related to the field of study:	M/NM_C.W3.+, M/NM_D.W5.+, M/NM_C.W7.+, C.U3.+, M/NM_C.W41.+, M/NM_K.7.+, D.U8.+, C.U2.+, M/NM_C.W22.+, M/NM_C.W42.+, C.U5.+, M/NM_A.W6.+, M/NM_C.W8.+, K.3.+, C.U1.+, D.U6.+, K.1.+, M/NM_C.W5.+, C.U4.+, KA7_KR1+, B.U10.+, M/NM_E.W39.+, M/NM_B.W18.+, M/NM_C.W2.+, M/NM_C.W1.+, M/NM_B.W14.+, M/NM_B.W13.+, M/NM_C.W4.+, M/NM_C.W6.+, M/NM_C.W11.+, M/NM_C.W9.+, M/NM_E.W37.+, M/NM_B.W15.+

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.

TEACHING FORMS AND METHODS:

Lecture(W1;):Theoretical lectures with the use of audiovisual presentations and problem methods (case method, didactic discussion, brainstorming, decision tree).
Seminar(W1;U1;K1;):Extending the subject of the lectures by presenting selected problems and clinical cases with active discussion in the seminar group.
Classes(W1;U1;K1;):Presentation of clinical cases with discussion in the exercise group.

FORM AND CONDITIONS OF VERIFYING LEARNING OUTCOMES:

Lecture (Colloquium test) - Lectures are held according to a fixed schedule. Attendance at lectures is obligatory and is checked at the beginning, during or at the end of the lecture. One excused absence is allowed. In the case of one excused absence, the student is obliged to prepare an essay on a given topic in the field of the lecture material. On the last day of the lecture block, a colloquium test is conducted on the thematic scope of the lectures. The colloquium test consists of 30 single-choice test questions. The condition for obtaining a pass is the correct answer to at least 20 out of 30 questions (66%). If the required number of correct answers is not obtained during the final colloquium test, the student is obliged to prepare an essay on a given topic from the scope of the lecture material. -
Seminar (Part in the discussion) - Part in the discussion - The student should theoretically be prepared for each class by mastering the necessary knowledge based on the literature or additional materials indicated by the teacher. The individual work of each student during the classes is assessed and credited by the teacher. Theoretical preparation and active participation in the classroom are taken into account when assessing and completing the course. -
Classes (Evaluation of the work and cooperation in the group) - Evaluation of the work and cooperation in the group - The condition for passing the exercises is attendance at all classes, active participation in the assessment and discussion of the presented clinical cases. -

BASIC LITERATURE:

1. Jorde L.B., Carey J.C., Bamshad M.J., *Genetyka medyczna*, Wyd. Elsevier, R. 2021
2. Connor J.M., Ferguson-Smith M.A., *Podstawy genetyki medycznej*, Wyd. Wydawnictwo Lekarskie PZWL, R. 1998
3. Friedman JM i wsp., *Genetyka*, Wyd. Urban i Partner, R. 2000
4. Korniszewski L., *Dziecko z zespołem wad wrodzonych. Diagnostyka dysmorfologiczna*, Wyd. Wydawnictwo Lekarskie PZWL, R. 2005
5. Kapelańska-Pręgowska J., *Prawne i bioetyczne aspekty testów genetycznych*, Wyd. Wolters Kluwer Polska, R. 2011
1. https://www.omim.org/
2. https://www.ncbi.nlm.nih.gov/books/NBK1116/

SUPPLEMENTARY LITERATURE:

1. Bennett R.L., *The Practical Guide to the Genetic Family History*, Wyd. Wiley-Blackwell, R. 2010
2. Darras B.T., Jones H.R., Ryan M.M., De Vivo D.C., *Neuromuscular Disorders of Infancy, Childhood, and Adolescence, Second Edition: A Clinician's Approach*, Wyd. Academic Press, R. 2014
3. Demkow U., Ploski R., *Applications for Next-Generation Sequencing*, Wyd. Academic Press, R. 2016
4. Firth H.V., Hurst J.A., *Oxford Desk Reference Clinical Genetics*, Wyd.

Oxford University Press, R. 2017
5. Jones K.L., Jones M.C., Del Campo M., *Smith's Recognizable Patterns of Human Malformation*, Wyd. Elsevier, R. 2013
6. Pevsner J., *Bioinformatics and Functional Genomics*, Wyd. Wiley-Blackwell, R. 2015
7. Rosenberg R.N., Pascual J.M., *Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease*, Wyd. Academic Press, R. 2015
8. Wood N., *Neurogenetics: A Guide for Clinicians*, Wyd. Cambridge University Press, R. 2012
9. Trygve T., *Handbook of Epigenetics: The New Molecular and Medical Genetics*, Wyd. Academic Press, R. 2011
10. Sadler T.W., *Langman's Medical Embryology*, Wyd. LWW, R. 2015
1. https://www.uniprot.org/
2. https://www.genecards.org/
3. https://www.face2gene.com/

Detailed description of ECTS credits awarded - part B

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ECTS: 2.00
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Clinical Genetics

The number of ECTS credits awarded consists of:

1. Contact hours with the academic teacher:

- participation in: Lecture	15.0 h
- participation in: Seminar	15.0 h
- participation in: Classes	10.0 h
- consultation	2.0
	Total: 42.0 h.

2. Independent work of a student:

Analysis of current literature on selected disease entities for later active participation in discussion during classes.	8.00 h

Total: 8.0 h

contact hours + independent work of a student Total: 50.0 h

1 ECTS credit = 25-30 h of an average student’s work, number of ECTS credit = 50.0 h : 25.0 h/ECTS = 2.00 ECTS on average: 2.0 ECTS

- including the number of ECTS credits for contact hours with the direct participation of an academic teacher: 0,00 ECTS points,

- including the number of ECTS credits for hours of independent work of a student: