SYLLABUS

The academic year when the cycle of instruction is commenced 2019-2025 [INT]

Module/course name:	**Clinical Genetics**	**Module code**	LK.3.C.006.1
Faculty:	Faculty of Medicine MUL		
Major:	Medical		
Specialty:			
Level of study:	I (Bachelor studies) ☐ II (Master studies) ☐ Integrated Master studies **X** Doctoral studies ☐		
Mode of study:	full-time **X** part-time (extramural) **X**		
Year of study:	I ☐ II ☐ III **X** IV ☐ V ☐ VI ☐	Semester:	1 ☐ 2 ☐ 3 ☐ 4 ☐ 5 **X** 6 ☐ 7 ☐ 8 ☐ 9 ☐ 10 ☐ 11 ☐ 12 ☐
Module/course type:	obligatory **X** elective ☐		
Language of instruction:	Polish ☐ English **X**		

Form of education	**Hours**
Lecture	**10**
Seminar	
Laboratory class	**25**
E-learning	
Practical class	
Internship	
Other	
TOTAL	
Student's work input (participation in class, preparation, evaluation, etc.)	**Student's hourly workload**
1. In class	**35**
2. Student's own work including: 1 Preparation for class 2 Preparation for partials and finals	**15**
Summary of the student's workload	**50**
ECTS points for module/course	**2**

Educational objectives:

After completing the lectures and exercises, the student should have current knowledge of clinical genetics, in particular: know the basic concepts of genetics, diagnosis and inheritance of monogenic, multifactorial and mitochondrial diseases, chromosomal structure, pathogenesis of chromosomal aberrations, rules of karyotype registration, recognition of the most common human syndromes chromosomal and developmental defects, know the basics of diagnosis of gene mutations and chromosomal diseases responsible for hereditary and acquired diseases, including neoplastic diseases, know different types of gender determination and gender inheritance rules, describe the principles of interaction and feedback genes, know the factors affecting the primary and secondary genetic balance of the population, analyze genetic flows and pedigrees of human traits and diseases, estimate the risk of disease in the offspring based on family predisposition and environmental factors, identify the indicated and prenatal, know the indications for cytogenetic and molecular tests, learn the basics of genetic counseling, the organization of genetic care in Poland, basic ethical problems in genetics, perspectives of clinical genetics development, gene therapy and therapy directed to specific diseases and be able to apply the acquired knowledge in practice.

The matrix of learning outcomes for module/ subject with reference to verification methods of the intended educational outcomes and forms of instruction:

Learning outcome code	A student who has obtained a credit for the module/course has the knowledge/skill to:	Methods of verifying the achievement of the intended learning outcomes:	Form of instruction * provide the symbol
C.W1	knows the basic concepts in genetics;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	Lecture/class
C.W2.	can describe the phenomena of gene feedback and cooperation;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.W3.	can describe normal human karyotype and various types of sex determination;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.W4.	can describe chromosome structure and molecular background of mutagenesis	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.W5.	knows the principles of inheritance of different number of characters, inheritance of quantitative characters, independent inheritance of characters and extranuclear inheritance;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	

C.W6.	knows genetic predispositions associated with human blood groups and serological conflict in respect to Rhesus factor;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.W7.	can describe autosomal and heterosomal aberrations causing diseases, including cancer, oncogenesis;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.W8.	knows factors affecting primary and secondary genetic equilibrium in a population;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.W9.	knows fundamentals of diagnosing gene and chromosomal mutations responsible for inherited and acquired diseases, including cancers;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.U1.	analyses genetic crosses and origins of human characteristics and diseases and assesses the risk of birth of a child having chromosomal aberrations;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.U2.	identifies indications for performing prenatal tests;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	

C.U3.	is capable of taking decision on the need to perform cytogenetic and molecular tests;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.U4.	performs morphometric measurements, analyses morphograms and records karyotypes of diseases;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	
C.U5.	assesses the risk for a given disease to appear in descendents, basing on family predispositions and influence of environmental factors;	written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) presentation	

EXAMPLES OF METHODS VERIFYING THE ACHIEVEMENT OF THE INTENDED LEARNING OUTCOMES:

In terms of knowledge: Oral exam (*non-standardized, standardized, traditional, problem-based*).
Written exam – the student produces/identifies answers)*essay, report; structured short-answer questions /SSQ/; multiple choice questions /MCQ/; multiple response questions /MRQ/; matching test; true/false test; open cloze test*)

In terms of skills: practical exam; Objective Structured Clinical Examination /OSCE/; Mini-CEX (mini – clinical examination); completion of a given assignment; project, presentation.

In terms of social competences:
A reflective essay; an extended observation by a supervisor/tutor; 360-degree assessment (feedback from teachers, peers, patients, other co-workers); self-assessment (portfolio included).

Course content: (use keywords referring to the content of each class following the intended learning outcomes):

Lectures:

1. Medical and clinical genetics. Types of genetic diseases.
2. Single gene defects (two parts)
3. Chromosomal abnormalities (two parts)
4. Multifactorial problems
5. Mitochondrial disorders
6. Population genetics
7. Genetics of cancer
8. Teratogenic problems

Seminar:

1. Introduction to clinical genetics.
Mendels law. Types of inheritance.

2. Single gene alteration I. (Quiz 1)
Autosomal dominant disorders: achondroplasia, Apert syndrome, Huntington disease, neurofibromatosis (NF-1, NF-2), Marfan syndrome, osteogenesis imperfecta.

3. Single gene alteration II. (Quiz 2)
Sex-linked disorders: Ehlers-Danlos syndrome, glucose-6-phosphate dehydrogenase deficiency (G6PD),

hemophilia A, hemophilia B; Duchenne muscular dystrophy, Becker muscular dystrophy.

4. Single gene alteration III. (Quiz 3)

Autosomal recessive disorders: albinism, cystic fibrosis, galactosemia, Gaucher disease, haemochromatosis; mucopolisaccharidoses, phenyloketonuria, sickle-cell anemia, Tay-Sachs disease, the thalassemias.

5. Multofactorial diseases. (Quiz 4)

Multifactorial diseases: Alzheimer disease, Parkinson disease, atherosclerosis, autism, diabetes mellitus, epilepsy, hypertension; neuronal tube defects, myocardial infarction, multiple sclerosis, schizophrenia, spina bifida, stroke.

6. Clinical symptoms of chromosomal syndromes I. (Quiz 5)

Autosomal aberrations (Down syndrome, translocation Down syndrome, Patau syndrome, Edward`s syndrome, cat`s cry syndrome, fragile X syndrome).

7. Clinical symptoms of chromosomal syndromes II. (Quiz 6)

Sex determination, sex chromosomes, sex-chromatin body. Sex-chromosomes anomalies (Turner syndrome, Klinefelter syndrome). Indications for karyotype examination.

8. Imprinting. (Quiz 7)

Genetic imprinting in human diseases. Prader-Willi, Angelman, Beckwith-Wiedemann, Russell-Silver syndromes. Imprinting and cancer.

9. Population genetics. (Quiz 8)

Hardy-Weinberg Law. Genetic pedigree. Identification of genetic disease risk.

Laboratory class:

1. Laboratory practice I. (Quiz 9)
   Cytogenetics.
2. Laboratory practice II. (Lab Quiz 1)
   Lymphocyte isolation.
3. Laboratory practice III. (Quiz 10; from topic 12)

(unnecessary delete)

Obligatory literature for lectures:

1. L.B. Jorde, J.C. Carey, M.J. Bamshad "Medical Genetics (4th edition)" MOSBY ELSEVIER, 2016;

2. "Human genetics: from molecules to medicine" C.P. Schaaf, J. Zschocke, L. Potocki - Lippincott Williams & Wolters Kluwer business, 2012 (firsth edition).

Complementary literature for lectures:

1. "GENETICS" Ronald W. Dudek – Lippincott Williams & Wolters Kluwer business, 2010. ISBN 978-0-7817-9994-2.

Obligatory literature for labs:

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Requirements for didactic aids (e.g. laboratory, multimedia projector, others…)

1.Laptop
2.Multimedia projector
3.Laboratory

Conditions for obtaining a credit for the subject:

- getting 60% of the points from the exercises
- getting 60% of points from the exam
- having a sufficient number of attendance (no more than 2 absences)

The name and address of the department/clinic where the course is taught (module/course); contact details (phone number/ email address):

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Names of the teacher/teachers conducting classes:

1. Prof. Janusz Kocki
2. Marcin Czop, PhD
3. Paulina Gil-Kulik, PhD
4. Alicja Petniak, PhD

Signature of the head of the department/clinic

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Dean's signature

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Date of submission: