| The acade   | mic year when the cy                     | SYLLABU                   |                           | enced 2019-2025 [INT                    | וי                               |  |  |  |  |  |  |  |  |
|---|--|---------------------------|---------------------------|---|----------------------------------|--|--|--|--|--|--|--|--|
| Module/course name:   | Medical Genetics                         | cie of moti det           | ion is comin              |   | LK.3.C.006                       |  |  |  |  |  |  |  |  |
| Faculty:  | Faculty of Medicine                      |                           |                           |   |                                  |  |  |  |  |  |  |  |  |
| Major:  | Medical                                  |                           |                           |   | 15-11-11                         |  |  |  |  |  |  |  |  |
| Specialty:  |  |                           |                           |   |                                  |  |  |  |  |  |  |  |  |
| Level of study:   | I (Bachelor studies) Doctoral studies    | II (Mast                  | er studies)               | Integrated Master st                    | udies X                          |  |  |  |  |  |  |  |  |
| Mode of study:  | full-time X part-tin                     | time (extramural) X       |                           |   |                                  |  |  |  |  |  |  |  |  |
| Year of study:  | I 🗆 II 🗆 III X IV 🗆                      | □ V□ VI □                 | Semester:                 | 1                                       | 6 7 8 8                          |  |  |  |  |  |  |  |  |
| Module/course type:   | obligatory X elect                       | tive 🗆                    |                           |   |                                  |  |  |  |  |  |  |  |  |
| Language of instruction:  | Polish   English                         | sh X                      |                           |   |                                  |  |  |  |  |  |  |  |  |
| Form of education   |  | Hours                     |                           |   |                                  |  |  |  |  |  |  |  |  |
| Lecture   |  | 10                        |                           |   |                                  |  |  |  |  |  |  |  |  |
| Seminar   |  |                           |                           |   |                                  |  |  |  |  |  |  |  |  |
| Laboratory class  |  | 25                        |                           |   |                                  |  |  |  |  |  |  |  |  |
| E-learning  |  |                           |                           |   |                                  |  |  |  |  |  |  |  |  |
| Practical class   |  |                           |                           |   |                                  |  |  |  |  |  |  |  |  |
| Internship  |  |                           | # - W                     |   |                                  |  |  |  |  |  |  |  |  |
| Other   |  |                           |                           |   |                                  |  |  |  |  |  |  |  |  |
| TOTAL   |  |                           |                           |   |                                  |  |  |  |  |  |  |  |  |
| Student's work input  (participation in class, preparation, eval  | luation, etc.)                           | Student's hourly workload |                           |   |                                  |  |  |  |  |  |  |  |  |
| 1. In class   |  | 35                        |                           |   |                                  |  |  |  |  |  |  |  |  |
| Student's own work included 1 Preparation for cla     Preparation for particles                                   | ss                                       | 15                        |                           |   |                                  |  |  |  |  |  |  |  |  |
| Summary of the student's wo   | orkload                                  | 50                        |                           |   |                                  |  |  |  |  |  |  |  |  |
| ECTS points for module/co   | ourse                                    | 2                         |                           |   |                                  |  |  |  |  |  |  |  |  |
| Educational objectives: The course consists of Chromosomal aberration Oncogenesis and Molecu with the Final Test. | s, Pathomechanism<br>lar methods. During | ns of genet<br>the course | ic diseases<br>students w | , Chromosomes, Imrite short quizzes. Tl | nmunogenetics,<br>he course ends |  |  |  |  |  |  |  |  |
| The matrix of learning out  | comes for module/ sul                    | ojeci with ref            | erence to ve              | rilication methods of t                 | ine intended                     |  |  |  |  |  |  |  |  |

educational outcomes and forms of instruction:

|                       |   |  | r   |
|-----------------------|---|--|---|
| 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 |
| B.W13.                | knows the function of nucleotides in a cell, primary and secondary structures of DNA and RNA and chromatin structure  | written exam (MCQ, matching test, true false test) short quiz (MCQ, true/false test, open questions) | Lecture/class                             |
|                       |   | presentation   |   |
|                       | knows the function of human genome, transcriptome and proteome and basic methods applied in their studies; describes the processes of replication, repair and | written exam (MCQ, matching test, true false test)   | Lecture/class                             |
| B.W14.                | recombination of DNA, transcription and translation and degradation of DNA, RNA and proteins; knows the concept of gene expression control                    | short quiz (MCQ,<br>true/false test, open<br>questions)  |   |
|                       |   | presentation   | ,   |
|                       | knows the basic concepts in genetics  | written exam (MCQ, matching test, true false test)   | Lecture/class                             |
| C.W1.                 |   | short quiz (MCQ,<br>true/false test, open<br>questions)  |   |
|                       |   | presentation   |   |
|                       | can describe the phenomena of gene feedback and cooperation   | written exam (MCQ, matching test, true false test)   | Lecture/class                             |
| C.W2.                 |   | short quiz (MCQ,<br>true/false test, open<br>questions)  |   |
|                       |   | presentation   |   |
|                       | can describe normal human karyotype and various types of sex determination  | written exam<br>(MCQ, matching<br>test, true false test)   | Lecture/class                             |
| C.W3.                 |   | short quiz (MCQ,<br>true/false test, open<br>questions)  |   |
|                       |   | presentation   |   |
|                       |   |  |   |

| -3  |       |  |  |               |
|-----|-------|--|--|---------------|
|     |       | can describe chromosome structure and molecular background of mutagenesis  | written exam (MCQ, matching test, true false test)       | Lecture/class |
| C   | .W4.  |  | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|     |       |  | presentation   |               |
|     |       | knows the principles of inheritance of different   | written exam (MCQ, matching                              | Lecture/class |
|     |       | number of characters, inheritance of quantitative characters, independent inheritance  | test, true false test)                                   |               |
| C.  | .W5.  | of characters and extranuclear inheritance   | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|     |       |  | presentation   |               |
|     |       | knows genetic predispositions associated with human blood groups and serological conflict in respect to Rhesus factor              | written exam (MCQ, matching test, true false test)       | Lecture/class |
| c.  | C.W6. |  | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|     |       |  | presentation   |               |
|     | C.W7. | can describe autosomal and heterosomal aberrations causing diseases, including cancer, oncogenesis                                 | written exam (MCQ, matching test, true false test)       | Lecture/class |
| C.' |       |  | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|     |       |  | presentation   |               |
|     |       | knows fundamentals of diagnosing gene and chromosomal mutations responsible for inherited and acquired diseases, including cancers | written exam<br>(MCQ, matching<br>test, true false test) | Lecture/class |
| C.1 | W9.   |  | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|     |       |  | presentation   |               |
|     |       | assesses benefits and risks resulting from presence in the ecosystem of genetically modified organisms(GMO)                        | written exam (MCQ, matching                              | Lecture/class |
|     |       |  | test, true false test)                                   |               |
| C.W | V10.  |  | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|     |       |  | presentation   |               |

|        | knows the consequences of exposing human organism to various chemical and biological agents and the rules of prevention                                   | written exam (MCQ, matching test, true false test)       | Lecture/class |
|--------|---|--|---------------|
| C.W15. |   | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|        |   | presentation   |               |
| C.W22. | can describe major histocompatibility complex   | written exam (MCQ, matching test, true false test)       | Lecture/class |
|        |   | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|        |   | presentation   |               |
| C.U1.  | analyses genetic crosses and origins of human characteristics<br>and diseases and assesses the risk of birth of a child having<br>chromosomal aberrations | written exam (MCQ, matching test, true false test)       | Lecture/class |
|        |   | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|        |   | presentation   |               |
|        | identifies indications for performing prenatal tests  | written exam (MCQ, matching test, true false test)       | Lecture/class |
| C.U2.  |   | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|        |   | presentation   |               |
|        | is capable of taking decision on the need to perform cytogenetic and molecular tests  | written exam (MCQ, matching test, true false test)       | Lecture/class |
| C.U3.  |   | short quiz (MCQ,<br>true/false test, open<br>questions)  |               |
|        |   | presentation   |               |
| C.U4.  | performs morphometric measurements, analyses morphograms and records karyotypes of diseases   | written exam<br>(MCQ, matching<br>test, true false test) | Lecture/class |
|        |   | short quiz (MCQ,<br>true/false test, open<br>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)      | Lecture/class |
|-------|---|---|---------------|
|       |   | short quiz (MCQ,<br>true/false test, open<br>questions) |               |
|       |   | presentation  |               |
| C.U6. | assesses environmental hazards and uses basic methods allowing to find the presence of harmful factors (biological and chemical) in the biosphere |   |               |

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

<u>In terms of knowledge:</u> 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 )

<u>In terms of skills:</u> 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. Chromosome analysis meiosis and mitosis, cell cultures, classical cytogenetics (banding techniques), molecular cytogenetics FISH, CGH, array CGH.
- 2. Immunogenetics.
- 3. Molecular methods in medical genetics.
- 4. Prenatal diagnostics aims, indications, noninvasive and invasive testing techniques, preimplantation testing.
- 5. Types of RNA. Role of non-coding RNA. MicroRNAs their biogenesis and function, miRNAs as prognostic and predictive factors.

# Laboratory class:

- LAB 1. INTRODUCTION TO GENETICS
- LAB 2. HUMAN GENOME
- LAB 3. POINT MUTATIONS AND POLYMORPHISMS
- LAB 4. PATHOMECHANISMS OF GENETIC DISEASES
- LAB 5. CHROMOSOMES AND THEIR ABERRATIONS
- LAB 6. CHROMOSOME ANALYSIS
- LAB 7. FROM GENES TO PROTEINS
- LAB 8. IMMUNOGENETICS
- LAB 9. Molecular methods PCR method and its variants.
- LAB 10. Molecular methods DNA sequencing.
- LAB 11. ONCOGENESIS PART I
- LAB 12. ONCOGENESIS PART II

# **Obligatory literature:**

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

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

### Complementary literature:

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

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

Laptop and multimedia projector.

# Conditions for obtaining a credit for the subject:

Presence during classes and lectures.

Students are obliged to be familiar with the issues covered by the lecture held during respective week and with the material to be discussed during classes according to the topic schedule.

To PASS the Final Test, the required final score is 60% of correctly answered questions out of all questions.

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

#### DEPARTMENT OF CANCER GENETICS WITH CYTOGENETIC LABORATORY

Address: RADZIWILLOWSKA STREET 11 (COLLEGIUM MEDICUM BUILDING),

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## Names of the author/authors of this syllabus:

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# Names of the teacher/teachers conducting classes:

Agata Filip, PhD Szymon Zmorzyński, PhD Sylwia Popek-Marciniec, PhD

| Signature of | f the hea | id of the | departm | ent/clinic |
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dr hab. n. med. Agata Filip

Date of submission:

| Dean's signature |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|------------------|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|--|
|                  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
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