Załącznik nr 5 do zarządzenia nr 189/2025

COURSE SPECIFICATION

Course code: 0912.4.LEK.C.Gen

Course title in Polish: Genetyka
Course title in English: Genetics

1. Placement of the course within the study programme

1.1. Field of study	Medicine
1.2. Mode of study	Full-time
1.3. Level of study	Uniform Master's study
1.4. Profile of study	General academic
1.5. Course specification author	Dr hab. prof. UJK Wioletta Adamus-Białek
1.6. Contact	wioletta.adamus-bialek@ujk.edu.pl

2. Generał characteristics of the course

2.1. Language of instruction	English
2.2. Entry requirements	Biology

3. Detailed characteristics of the course

3.1. Type of classes	Lectures (L) – 20h, including 5h of e-learning, Classes (C) – 20h, Laboratories (Lab) – 10h
3.2. Place of course delivery	traditional classes in the UJK teaching room
3.3. Form of assessment	Lecture: exam (4th semester), Classes: credit with grade, Laboratories: credit with grade.
3.4. Teaching methods	Lecture (including e-learning): informative lecture, conversational lecture, problem-based lecture Classes: Subject-specific exercises, case studies Laboratories: Laboratory exercises, practical exercises, measurement, demonstration with explanation, demonstration with instruction
3.5.a. Basic reading list	1.Medical Genetics, by Lynn B. Jorde PhD, John C. Carey MD MPH, Michael J. Bamshad MD, Elsevier, last edition 2.Essential Medical Genetics (with FREE Desktop Edition) 6/e, Tobias,Connor,Ferguson-Smith, WILEY, last edition
3.5.b. Supplementary reading list	3.Color Atlas of Genetics (FLEXIBOOK) by Eberhard Passarge 4. DeVita, Hellman and Rosenberg's Cancer: Principles & Practice of Oncology, last edition

4. Objectives, content and learning outcomes

4.1. Course objectives (taking into account the form of classes)

Lectures:

C1 Familiarizing students with the mechanisms of congenital defects and the hereditary basis of diseases and disorders

C2 Presenting hereditary predispositions to oncogenesis

C3 Classifying major genetic diseases

(including e-learning)

C4 Introducing the elements of genetic counseling
C5 Introducing ethical principles in genetic research

Classes:

C1. Presenting the principles of conducting a patient interview, constructing and analyzing a pedigree, and the fundamental rules of trait inheritance.
C2. Introducing the basics of dysmorphology, morphometric measurements, and features suggestive of a genetic basis for diseases.
C3. Presenting the fundamentals of cytogenetics, chromosomal aberrations, methods for their detection, and the conventions for recording and interpreting karyotypes.
C4. Providing knowledge on the application of molecular biology methods in genetic diagnostics, GWAS analysis, and the interpretation of genetic variants and markers.
C5. Presenting the genetic basis for the inheritance of blood groups, Rh incompatibility, and the significance of the HLA system in transplantology.
C6. Discussing factors that influence the genetic equilibrium of a population, as well as the principles for assessing genetic risk based on pedigrees and genetic test results.

Laboratory

C1. Presenting the application of the FISH technique in molecular diagnostics, including the preparation and interpretation of tests using probes employed in routine genetic diagnostics.
C2. Familiarization with the stages of preparing NGS libraries for microbiome analysis, including DNA isolation and quality assessment, as well as the amplification of specific regions of the 16S rRNA gene.
C3. Imparting practical skills for the purification, normalization, and indexing of NGS libraries, performing quality control, and preparing them for sequencing.
C4. Demonstrating practical skills in performing and interpreting selected genetic diagnostic methods.

4.2. Course content (taking into count the form of classes)

Lectures:

Mechanisms of development of congenital defects and hereditary determinants of diseases and disorders, including human sex determination. Genetic causes of hereditary predisposition to cancer. Genetic determinants of the most common monogenic, multigenic and multifactorial diseases, basic chromosome aberration syndromes, syndromes caused by genomic rearrangements, polymorphisms, epigenetic and post-transcriptional changes, including selected rare diseases. Causes, symptoms, principles of diagnosis and therapeutic procedures in the most common genetic diseases in children and adults;
(e-learning): Familiarization with the elements of genetic counseling and ethical principles in genetics

Classes:

Preparation for an interview with a patient in order to construct and analyze a pedigree. Principles of inheritance of different numbers of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of extranuclear genetic information. Basics of dysmorphology - morphometric measurements and characterization of dysmorphic features and other clinical and pedigree features suggesting the genetic basis of diseases. Basics of cytogenetics, including chromosome aberrations and methods of their detection, principles of writing and reading carytype. Molecular biology in genetic diagnostics and basic indications for their use, including detection of

genetic drug resistance of cancer cells in order to individualize pharmacotherapy. Causes and principles of diagnosis in the most common genetic diseases in children and adults. Analysis of genome-wide association studies (GWAS) and identification of genetic variants associated with predisposition to multifactorial diseases, including variants with low penetrance and without confirmed pathogenic status. Assessment of the diagnostic and clinical value of genetic markers and interpretation of results in the analysis of clinical cases. Genetic determinants of human blood groups. Inheritance of blood group antigens, with particular emphasis on the Rh system. Mechanisms and importance of Rh serological conflict in clinical practice. Genetic basis of transplantology. Inheritance and the importance of the HLA histocompatibility system in the selection of donors and recipients of organs and tissues. Factors influencing the primary and secondary genetic balance of the population. Determining genetic risk based on pedigree and genetic test results.

Laboratory:

Application of the FISH technique in molecular diagnostics. Principles of material preparation and the course of FISH testing on the example of assessing HER2 gene amplification in tumor material. Interpretation of results in accordance with current diagnostic guidelines. Preparation of NGS libraries for microbiome analysis by sequencing the 16S rRNA region: isolation and assessment of the quality of genetic material, amplification of selected regions of the 16S rRNA gene. purification and normalization of libraries, indexing of samples, quality control of libraries and preparation of material for sequencing.

4.3. Course learning outcomes

Course learning outcomes (code)	Upon completion of the course, the student	Alignment with programme learning outcomes
	in the area of knowledge knows and understands:	
W01	Normal human karyotype and various types of sex determination;	C.W1.
W02	Genetic causes of hereditary predisposition to cancer;	C.W2.
W03	The principles of inheritance, inheritance of quantitative traits, independent inheritance of traits and inheritance of extranuclear genetic information;	C.W3.
W04	The genetics of blood groups and serological conflict in Rh system;	C.W4.
W05	Genetic determinants of the most common monogenic and polygenic and multifactorial diseases, basic chromosome aberration syndromes, syndromes caused by genomic rearrangements, polymorphisms, epigenetic and post-transcriptional changes;	C.W5.
W06	The factors affecting primary and secondary genetic balance of the population;	C.W6.
W07	Genetic determinants of congenital malformations and selected rare diseases and the possibility of their prevention;	C.W7.
W08	Genetic diagnostic methods and basic indications for their use;	C.W8.
W09	Genetic mechanisms of drug resistance acquisition by microorganisms and tumor cells;	C.W9.
W10	The genetic basis for selection of the donor and recipient and the basics of the immunology of transplantation;	C.W22.
W11	Causes, symptoms, principles of diagnosing and therapeutic management in the most common genetically determined diseases in children and adults;	E.W36.
	in the area of skills knows how to:	

U01	Plot and analyze pedigrees and identify clinical and pedigree features suggesting the genetic basis of diseases;	C.U1.
U02	Make a decision on the need to perform cytogenetic and molecular tests;	C.U2.
U03	Read basic genetic test results, including karyotypes;	CU3.
U04	Determine genetic risk based on pedigree and genetic test results in the case of chromosomal aberrations, genomic rearrangements, single-gene and multifactorial diseases;	C.U4.

in the area of social competences is able to:

K01	Recognize his/her own limitations and self-evaluate educational deficiencies and needs;	K.S5.
K02	Use reliable information sources;	K.S7.
K03	Conclude on the basis of own surveys and observations;	K.S8.
K04	Introduce rules of social conduct and teamwork to the group of specialists, including specialists form other medical professions also in the multicultural and multinational environment;	K.S9.
K05	Give opinions concerning various aspects of professional activity;	K.S10.
K06	Take responsibility for own decisions made during professional activities including own safety and safety of other people.	K.S11.

4.4. Assessment methods for learning outcomes

Assessment method (+/-)

Course learning outcomes (code)	Oral/written/ practical examinaion /other (specify)*	Test*	Project*	In-class activity*	Independent study*	Group work*	Other (specify)*

Type of classes

1: / 2:	L	C	Lab	L	C	Lab	L	C	Lab	L	C	Lab	L	C	Lab	L	C	Lab	L	C	Lab
W01	+	+			+																
W02	+	+			+																
W03	+																				
W04		+			+																
W05	+	+			+																
W06		+																			
W07		+			+																
W08	+	+			+																
W09		+			+																
W10		+			+																
W11		+			+																
U01		+			+																
U02		+	+		+										+						
U03		+	+		+										+						
U04		+			+																
K01	+	+	+		+										+						
K02	+	+	+		+										+						
K03	+	+	+		+										+						
K04	+	+	+		+										+						

K05	+	+	+		+										+					
K06	+	+	+		+										+					

Annotation. 1: form of classes; 2: learning outcomes

4.5. Assessment criteria for learning outcomes

Type of classes:

LECTURE (L) (including classes conducted with the use of distance learning methods and techniques)

Grade	Assessment criteria
3,0	Obtaining 61-68% score on the test verifying the knowledge and skills provided for in the curriculum
3,5	Obtaining 69-76% score on the test verifying the knowledge and skills provided for in the curriculum
4,0	Obtaining 77-84% score on the test verifying the knowledge and skills provided for in the curriculum
4,5	Obtaining 85-92% score on the test verifying the knowledge and skills provided for in the curriculum
5,0	Obtaining 93-100% score on the test verifying the knowledge and skills provided for in the curriculum

Type of classes:

CLASSES (C) (including classes conducted with the use of distance learning methods and techniques)

Grade	Assessment criteria
3,0	Obtaining 61-68% score on the test verifying the knowledge and skills provided for in the curriculum
3,5	Obtaining 69-76% score on the test verifying the knowledge and skills provided for in the curriculum
4,0	Obtaining 77-84% score on the test verifying the knowledge and skills provided for in the curriculum
4,5	Obtaining 85-92% score on the test verifying the knowledge and skills provided for in the curriculum
5,0	Obtaining 93-100% score on the test verifying the knowledge and skills provided for in the curriculum

Type of classes:

LABORATORIES (Lab) (including classes conducted with the use of distance learning methods and techniques)

Grade	Passing 61-68% of the requirements specified by the tutor related to the preparation of the laboratory report
3,0	Passing 69-76% of the requirements specified by the tutor related to the preparation of the laboratory report
3,5	Passing 77-84% of the requirements specified by the tutor related to the preparation of the laboratory report
4,0	Passing 85-92% of the requirements specified by the tutor related to the preparation of the laboratory report
4,5	Passing 93-100% of the requirements specified by the tutor related to the preparation of the laboratory report
5,0	Passing 61-68% of the requirements specified by the tutor related to the preparation of the laboratory report

5. ECTS credit allocation – student workload

Category	Student workload (full-time mode)	Student workload (part-time mode)
NUMBER OF HOURS WITH THE DIRECT PARTICIPATION OF THE TEACHER (CONTACT HOURS)	**50**	
Participation in lectures*	15	
Participation in seminars/laboratories*	30	
Other (specify) e.g. classes conducted with the use of distance learning methods and techniques*	5	
INDEPENDENT STUDY (NON-CONTACT HOURS)	**45**	

Preparation for lectures*	15	
Preparation for seminars/laboratories*	30	
Preparation for examination/test*		
Gathering materials for project, online research*		
Preparation of a multimedia presentation*		
Other (specify)*		
TOTAL HOURS	**95**	
ECTS CREDITS for the course	**4**	

*** delete as appropriate**

Approved for implementation (date and legible signatures of course instructors in the given academic year):

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