

DESCRIPTION OF THE COURSE OF STUDY

Course code	0912-7LEK-C3.1-G	
Name of the course in	Polish	Genetyka
	English	Genetics

1. LOCATION OF THE COURSE OF STUDY WITHIN THE SYSTEM OF STUDIES

1.1. Field of study	Medicine
1.2. Mode of study	Full-time
1.3. Level of study	Uniform master's studies
1.4. Profile of study*	General academic
1.5. Person preparing the course description	Dr Wioletta Adamus-Bialek
1.6. Contact	

2. GENERAL CHARACTERISTICS OF THE COURSE OF STUDY

2.1. Language of instruction	English
2.2. Prerequisites*	Biology

3. DETAILED CHARACTERISTICS OF THE COURSE OF STUDY

3.1. Form of classes	LECTURE 15 hours.; CLASSES: 30 hours	
3.2. Place of classes	Courses in the teaching rooms of the UJK,	
3.3. Form of assessment	LECTURE – L, Cl. Credit with grade	
3.4. Teaching methods	Classical and Conversational lecture, discussion, classes	
3.5. Bibliography	Required reading	1. Medical Genetics, by Lynn B. Jorde PhD (Author), John C. Carey MD MPH (Author), Michael J. Bamshad MD (Author) 2. Essential Medical Genetics (with FREE Desktop Edition) 6/e, Tobias, Connor, Ferguson-Smith, WILEY, 2011
	Further reading	3. Color Atlas of Genetics (FLEXIBOOK) by Eberhard Passarge (Author) 4. DeVita, Hellman and Rosenberg's Cancer: Principles & Practice of Oncology, 10 th Ed.

4. OBJECTIVES, SYLLABUS CONTENT AND INTENDED LEARNING OUTCOMES

4.1. Course objectives (including form of classes)

Lectures:

- O1. Introduction to the basic knowledge in the field of genetics and its role in human life and health.
- O2. Acquiring the ability to use the nomenclature of medical genetics.
- O3. Acquiring the knowledge in the field of genetic counseling, gathering genetic history.
- O4. Shaping the correct physician's attitude in transmitting genetic information to the patient and his family.
- O5. Acquainting with genetic determinants of selected diseases with their basic clinical characteristics.

Classes:

- O1. Acquiring the ability to determine the types of inheritance, classification of birth defects and pedigree construction.
- O2. Acquiring skills in the use of diagnostic tests and their correct interpretation.
- O3. The ability to use basic knowledge in the analysis of inheritance and diagnosis of genetic diseases.
- O4. Understanding the mechanisms regulating DNA metabolism, its dysfunction and role in the development of genetic diseases

4.2. Detailed syllabus (including form of classes)

Lectures:

The cognitive values of the human genome in medical practice. Basic concepts in the field of genetics. Metabolism of DNA (replication, repair, recombination of DNA, transcription, translation, degradation of DNA and RNA, regulation of gene expression). The importance of GMO in medicine. Principles and methods of genetic counseling. Bioethics in genetics. Monogenic disorders. Syndrome with chromosomal instability. Mitochondrial disorders. Epigenetic, multifactorial diseases. Principles of inheriting predisposition to tumors. Molecular analysis

of DNA and RNA in detecting inherited predisposition to tumors, genetic mechanisms of drug resistance acquisition by tumor cells. Basic directions of gene therapy development in specific inherited diseases

Classes:

Rules and drawing pedigrees. Types of inheritance - determination based on inheritance. Basics of dysmorphology. Cytogenetics and analysis of karyotypes. Determination of sex and inheritance of blood groups in humans. Molecular basis of mutagenesis, DNA sequence analysis, searching for mutations/polymorphisms and evaluation of their pathogenicity. Analysis of genetic crosses and pedigrees of human traits and diseases, risk of a child's birth with chromosomal aberrations, estimation of the risk of a given disease appearing in the offspring based on family predisposition and the environmental factors influence. Probability calculations in pedigrees. The phenomenon of gene linkage and gene interaction. Multigene diseases: association analysis, relative risk, odds ratio. Principles of inheritance of various number of traits, inheritance of quantitative traits, independent inheritance of features and inheritance of non-nuclear genetic information. Genetic balance in the population.

4.3 Intended learning outcomes

Code	A student, who passed the course	Relation to learning outcomes
within the scope of KNOWLEDGE the graduate knows and understands:		
W1.	the functions of the human genome, transcriptome and proteome and basic methods used in their study; describes the processes of replication, repair and recombination of DNA, transcription and translation and degradation of DNA, RNA; the concept of the regulation of gene expression;	B.W14.
W2.	the basic concepts of genetics;	C.W1.
W3.	the phenomenon of linkage and interaction of genes;	C.W2.
W4.	normal human karyotype and various types of sex determination	C.W3.
W5.	the structure of chromosomes and the molecular mechanisms of mutagenesis;	C.W4.
W6.	the principles of inheritance, inheritance of quantitative traits, independent inheritance of traits and inheritance of extranuclear genetic information;	C.W5.
W7.	the genetics of blood groups and serological conflict in Rh system;	C.W6.
W8.	the aberrations of autosomes and heterosomes causing diseases, including cancer oncogenesis;	C.W7.
W9.	the factors affecting primary and secondary genetic balance of the population	C.W8.
W10.	the foundation for the diagnosis of gene and chromosome mutations responsible for hereditary and acquired diseases, including cancer;	C.W9.
W11.	benefits and risks arising from the presence in the ecosystem of genetically modified organisms (GMOs);	C.W10.
W12.	genetic mechanisms, the acquisition of drug resistance by tumor cells;	C.W11.
within the scope of ABILITIES the graduate knows how to:		
U1.	analyse genetic cross, pedigree qualities and human diseases as well as the estimated risk of having a child with chromosomal aberrations;	C.U1
U2.	identify indications for prenatal diagnosis;	C.U2.
U3.	make a decision on the need to perform cytogenetic and molecular tests	C.U3.
U4.	make morphometric measurements, analyzes the developmental profile and records the diseases' karyotypes;	C.U4.
U5.	assesse the risk of disclosure of a particular disease in the offspring based on family predisposition and the influence of environmental factors;	C.U5.

4.4. Methods of assessment of the intended learning outcomes																					
Teaching outcomes (code)	Method of assessment (+/-)																				
	Exam oral/written*			Test*			Project*			Effort in class*			Self-study*			Group work*			Others*		
	Form of classes			Form of classes			Form of classes			Form of classes			Form of classes			Form of classes			Form of classes		
	L	C	...	L	C	...	L	C	...	L	C	...	L	C	...	L	C	...	L	C	...
W1; W2; W3; W5; W7; W8; W9; W10; W11; W13	+												+								
... W4; W6; W12; W13; U1 – U5					+								+								

*delete as appropriate

4.5. Criteria of assessment of the intended learning outcomes		
Form of classes	Grade	Criterion of assessment
lecture (L)	3	61% - 68% Learning programme content on the basic level, replies chaotic, leading questions necessary.
	3,5	69%-76% Learning programme content on the basic level, answers systematized, requires assistance from the teacher.
	4	77%-84% Mastering course content at the primary level, response systematized, independent. Solving problems in typical situations.
	4,5	85%-92% The scope of presented knowledge exceeds the basic level based on the supplementary literature provided. Solving of problems in new complex situations
	5	93%-100% The scope of presented knowledge exceeds the basic level based on independently acquired scientific sources of information.
classes (C)*	3	61% - 68% Learning programme content on the basic level, replies chaotic, leading questions necessary.
	3,5	69%-76% Learning programme content on the basic level, answers systematized, requires assistance from the teacher.
	4	77%-84% Mastering course content at the primary level, response systematized, independent. Solving problems in typical situations.
	4,5	85%-92% The scope of presented knowledge exceeds the basic level based on the supplementary literature provided. Solving of problems in new complex situations
	5	93%-100% The scope of presented knowledge exceeds the basic level based on independently acquired scientific sources of information.

Conditions for obtaining credit.

The prerequisite is to obtain credits for all classes: lectures – mandatory presence, classes – presence in all classes and an active participation in the classes according to the schedule. In case of excused absence – the obligation of making up classes after consultation with an assistant professor.

The exam in the form of a written test with a grade.

5. BALANCE OF ECTS CREDITS – STUDENT’S WORK INPUT

Category	Student's workload
	Full-time studies
<i>NUMBER OF HOURS WITH THE DIRECT PARTICIPATION OF THE TEACHER /CONTACT HOURS/</i>	45
<i>Participation in lectures*</i>	15
<i>Participation in classes, seminars, laboratories*</i>	30
<i>Preparation in the exam/ final test*</i>	
<i>Others*</i>	
<i>INDEPENDENT WORK OF THE STUDENT/NON-CONTACT HOURS/</i>	55
<i>Preparation for the lecture*</i>	5
<i>Preparation for the classes, seminars, laboratories*</i>	35
<i>Preparation for the exam/test*</i>	15
<i>Gathering materials for the project/Internet query*</i>	
<i>Preparation of multimedia presentation</i>	
<i>Others (please specify e.g. e-learning)*</i>	
TOTAL NUMBER OF HOURS	100
ECTS credits for the course of study	4

**delete as appropriate*

Accepted for execution (date and legible signatures of the teachers running the course in the given academic year)

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