

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. Specialization*	Lack
1.6. Unit running the course of study	Faculty of Medicine and Health Sciences
1.7. Person/s preparing the course description	dr hab. n. med. Stanisław Gózdź, prof.UJK
1.8. Person responsible for the course of study	dr n.biol. Wioletta Adamus - Białek
1.9. Contact	dr n. biol.Wioletta Adamus – Białek; dr n. biol. Michał Majchrzak

2. GENERAL CHARACTERISTICS OF THE COURSE OF STUDY

2.1. Affiliation with the module	Preclinical studiem
2.2. Language of instruction	English
2.3. Semesters in which the course of study is offered	3 rd semester
2.4. 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, Faculty of Molecular Diagnostics, Genetic Counseling ŚCO
3.3. Form of assessment	LECTURE – L, CI. Credit with grade
3.4. Teaching methods	Conversational lecture, discussion, a case study in natural condition
3.5. Bibliography	Required reading
	Further 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
 3. Color Atlas of Genetics (FLEXIBOOK) by Eberhard Passarge (Author)
 4. DeVita, Hellman and Rosenberg's Cancer: Principles & Practice of Oncology, 10th Ed.

4. OBJECTIVES, SYLLABUS CONTENT AND INTENDED TEACHING OUTCOMES

4.1. Course objectives

- C1 Transferring the basic knowledge of genetics- types of inheritance, classification of birth defects, genetics counseling.
 C2 Acquiring the skills of gathering genetic interview and consulting pedigrees, using the diagnostic tests as well as their correct interpretation.
 C3 Forming a proper doctor attitude in the transfer of genetic information to the patient and their family.

4.2. Detailed syllabus

Lectures

1. Cognitive value of the human genome in medical practice. Basic genetic concepts; 3 hours.
2. Monogenic diseases. The principles of genetic counseling; 3 hours.
3. The set with instability of chromosomes. Mitochondrial disease; 2 hours.
4. Epigenetic disease, multifactorial; 1 hour.
5. Bioethics in genetics; 2 hours.
6. Ways of developing genetic counseling; 2 hours.
7. Principles of inheritance predisposition to cancer. Molecular analysis DNA and RNA in detection of genetic predisposition to cancer.

Classes

1. Principles of writing pedigree. Drawing pedigrees; 3 hours.
2. Types of inheritance- determination on the basis of inheritance; 3 hours.
3. Probability, condition probability, genetic risk; 3 hours.
4. Calculations of probability in the pedigrees; 3 hours.
5. Connections analysis; 3 hours.
6. Polygenic diseases: association analysis, relative risk, odds ratio; 3 hours.
7. The basis of dysmorphology; 3 hours.
8. Molecular genetics. Sequencing analysis of DNA, the search for mutations, polymorphism and assess of their pathogenicity; 3 hours.
9. Cytogenetics. Analysis of karyotypes. 3 hours.
10. Credit. 3 hours.

4.3. Education outcomes in the discipline

Code	A student, who passed the course	Relation to teaching outcomes
within the scope of KNOWLEDGE:		
W1.	knows 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 and proteins; knows the concept of the regulation of gene expression;	B.W14. C.W1.
W2.	knows the basic concepts of genetics;	C.W1.
W3.	describes the phenomenon of coupling and interaction of genes;	C.W2.
W4.	describes normal human karyotype and various types of sex determination	C.W3.
W5.	describes the structure of chromosomes and the molecular mechanisms of mutagenesis;	C.W4.
W6.	knows the principles of inheritance, inheritance of quantitative traits, independent inheritance of traits and inheritance of extranuclear genetic information;	C.W5.
W7.	knows the genetics of blood groups and serological conflict in Rh system;	C.W6.
W8.	describes the aberrations of autosomes and heterosomes causing diseases, including cancer oncogenesis;	C.W7.
W9.	knows the factors affecting primary and secondary genetic balance of the population	C.W8.
W10.	knows the foundation for the diagnosis of gene and chromosome mutations responsible for hereditary and acquired diseases, including cancer;	C.W9.
W11.	determines benefits and risks arising from the presence in the ecosystem of genetically modified organisms (GMOs);	C.W10.
W12.	knows genetic mechanisms, the acquisition of drug resistance by microorganisms and tumor cells;	C.W11.
W13.	knows and understand the causes, symptoms, principles of diagnosis and therapeutic management of the most common hereditary diseases;	E.W35.

within the scope of ABILITIES:		
U1.	analyses genetic crossing over, pedigree qualities and human diseases as well as the estimated risk of having a child with chromosomal aberrations;	C.U1
U2.	identifies indications for prenatal diagnosis;	C.U2.
U3.	makes a decision on the need to perform cytogenetic and molecular tests	C.U3.
U4.	makes morphometric measurements, analyzes the developmental profile and records the diseases' karyotypes;	C.U4.
U5.	assesses 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 teaching outcomes

Teaching outcomes (code)	Method of assessment (+/-)																				
	Exam /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					
	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					+																

4.5. Criteria of assessment of the intended teaching 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.

- **Thresholds are valid from 2018/ 2019 academic year**

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*</i>	
TOTAL NUMBER OF HOURS	100
ECTS credits for the course of study	4

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

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