

DESCRIPTION OF THE COURSE OF STUDY

Course code	0511-2BIO-BC11-GC	
Name of the course in	Polish	Genetyka człowieka
	English	Human genetics

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

1.1. Field of study	Biology
1.2. Mode of study	full-time studies
1.3. Level of study	second-degree studies
1.4. Profile of study*	general-academic
1.5. Person/s preparing the course description	dr hab Artur Kowalik, dr Ernest Skowron
1.6. Contact	artur.kowalik@ujk.edu.pl, ernest.skowron@ujk.edu.pl

2. GENERAL CHARACTERISTICS OF THE COURSE OF STUDY

2.1. Language of instruction	English
2.2. Prerequisites*	Knowledge of the subjects Biochemistry and Genetics, at the undergraduate level

3. DETAILED CHARACTERISTICS OF THE COURSE OF STUDY

3.1. Form of classes	lectures, laboratory, (including e-learning)	
3.2. Place of classes	Classes in the teaching facilities of UJK	
3.3. Form of assessment	graded credit (TEST),	
3.4. Teaching methods	Lecture and discussions, laboratory courses	
3.5. Bibliography	Required reading	1. Brown T.A., Genomy, PWN, Warszawa, 2019. (Genomes 4 4th Edition English) 2. Connor M., Ferguson-Smith M., Tobias E., Genetyka medyczna, PZWL, Warszawa, 2013 (Essential Medical Genetics, Includes Desktop Edition, 6th Edition English) 3. Węgleński P. (red.), Genetyka molekularna, PWN, Warszawa, 2012.
	Further reading	1. Bal J. (red.), Biologia molekularna w medycynie. Elementy genetyki klinicznej. PWN, Warszawa, 2011. 2. PubMed

4. OBJECTIVES, SYLLABUS CONTENT AND INTENDED LEARNING OUTCOMES

4.1. Course objectives (including form of classes) C1. Learning about the structures responsible for the organization of genetic material (lecture). C2. Learning about the determinants of genetic variation (lecture). C3. Learning about the methods of analysis of genetic material (laboratory exercises) C4. Learning about genetically determined diseases and disorders of body functions and determining the frequency of their occurrence in the population (laboratory exercises)	4.2. Detailed syllabus (including form of classes) Lectures Lecture content: - Organization of the human genome: nuclear genome, mitochondrial genome. Human karyotype (structure and types of chromosomes). Variability of organisms (genotype/phenotype). Chromosome aberrations/Mutations and DNA polymorphisms. Genetic variability of populations (rate of change of nucleotide sequences, rate of evolutionary change). Population genetic diversity (balanced/unbalanced population (Wright statistic, polymorphism information index, nucleotide diversity). Demographic effects (founder, bottleneck). Genealogy of sequences (coalescence). Evolutionary models (regional continuity, recent common ancestor). Evolution of continental populations (age assessment, demographic expansion). Genetic counseling (genotypic diagnosis, phenotypic diagnosis). Assessment of the risk of occurrence/recurrence of disease. Genetic determinants of metabolic diseases, cancer (genes associated with the cancer process). Aspects of human genome research (ethical, medical, moral, political). Legislative standards. laboratory Organization of the human genome. Human karyotype (structure and types of chromosomes, basics of inheritance - scientific, chromosomal, chemical). Genetic diversity of the population (Hardy-Weinberg principle - assumptions, calculations in the field of population genetics.). Demographic effects (founder, bottleneck). Genealogy. Chromosomal aberrations. DNA mutations and polymorphisms (application of restriction fragment length polymorphism in genetic analyzes. Single nucleotide polymorphism of SNP). Genetic determinants of metabolic and neoplastic diseases (genes related to the neoplastic process, analysis of mitochondrial diseases). Assessment of the risk of disease occurrence / recurrence (pedigree analysis). Aspects of the study of the human genome (medical).
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4.3 Intended learning outcomes

Code	A student, who passed the course	Relation to learning outcomes
within the scope of KNOWLEDGE:		
W01	Knows the organization and principles of the genetic apparatus	BIO2A_W07
W02	Knows the basics of genetic variation in organisms	BIO2A_W07
W03	Knows the evolutionary and human population variability	BIO2A_W09
W04	Has knowledge of the dilemmas and ethical and legal determinants of genetics	BIO2A_W09
within the scope of ABILITIES:		
U01	Can practically use techniques and research tools, including advanced information and communication techniques in the field of analysis of genetic variability of organisms, and can interpret the obtained results	BIO2A_U02
within the scope of SOCIAL COMPETENCE:		
K01	Updates his knowledge of human genetic processes	BIO2A_K01

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* e.g. standardized test used in e-learning		
	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	...
W01				+	+					-	+					-	+				
W02				+	+					-	+					-	+				
W03				+	+					-	+					-	+				
W04				+	+					-	+					-	+				
U01				+	+					-	+					-	+				
K01				-	-					-	+					-	+				

*delete as appropriate

4.5. Criteria of assessment of the intended learning outcomes

Form of classes	Grade	Criterion of assessment
lecture (L) (including e-learning)	3	Obtaining 51-65% of the total number of points possible to obtain
	3,5	Obtaining 66-75% of the total number of points possible to obtain
	4	Obtaining 76-85% of the total number of points possible to obtain
	4,5	Obtaining 86-95% of the total number of points possible to obtain
	5	Obtaining 96-100% of the total number of points possible to obtain
laboratory (C)* (including e-learning)	3	Obtaining 51-65% of the total number of points possible to obtain
	3,5	Obtaining 66-75% of the total number of points possible to obtain
	4	Obtaining 76-85% of the total number of points possible to obtain
	4,5	Obtaining 86-95% of the total number of points possible to obtain
	5	Obtaining 96-100% of the total number of points possible to obtain

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

Category	Student's workload	
	Full-time studies	Extramural studies
NUMBER OF HOURS WITH THE DIRECT PARTICIPATION OF THE TEACHER /CONTACT HOURS/	40	
Participation in lectures*	20	
Participation in laboratories*	20	
Preparation in the final test*		
Others (please specify e.g. e-learning)*		
INDEPENDENT WORK OF THE STUDENT/NON-CONTACT HOURS/	60	
Preparation for the lecture*	10	
Preparation for the, laboratories*	15	
Preparation for the test*	30	
Gathering materials for the Internet query*	5	
Preparation of multimedia presentation		
Others *		
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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