

Course description

Part 1

General information about the course		
1. Major of study: medicine	2. Study level: unified MSc	
	3. Form of study: intramural	
4. Year: III	5. Semester: VI	
6. Course name: Genetics II		
7. Course status: required		
8. Course contents and assigned learning outcomes		
Bases of clinical genetics Pedigree construction, recognition of inheritance patterns on the basis of pedigree analysis. Division, clinical picture, the risk of recurrence of genetic disorders. Indications, rules, schema in genetic counseling. Prenatal diagnosis . Chromosomal abnormalities. Numerical and structural chromosomal aberrations and contiguous gene syndromes. Autosomal dominant, autosomal recessive, and X-linked diseases. Congenital abnormalities. Incidence, definitions, etiology and classification of birth defects.		
Learning outcomes / reference to learning outcomes indicated in the standards		
For knowledge – student knows and understands: C.W7; C.W9; C.W41; C.W42; E.W3.10; E.W25; E.W37		
For skills student can do: B.U10; C.U3; C.U5		
For social competencies student is ready to: percept and to recognize personal limitations and to accomplish self-evaluation of his/her own deficits and educational needs; to benefit from objective sources of information		
9. Number of hours for the course		60
10. Number of ECTS points for the course		4
11. Methods of verification and evaluation of learning outcomes		
Learning outcomes	Methods of verification	Methods of evaluation*
Knowledge	Written evaluation – open questions Grade credit – MCQ Test of Choice Yes/No Test of matching answers Short structural questions	* Exam - MCQ
Skills	Worksheets Observation Multimedial presentation	* Passing of personally prepared multimedial presentation * correct completion of the worksheets
Competencies	Observation	* Observation

* The following evaluation system has been assumed:

Very good (5,0) – the assumed learning outcomes have been achieved and significantly exceed the required level

Better than good (4,5) – the assumed learning outcomes have been achieved and slightly exceed the required level

Good (4,0) – the assumed learning outcomes have been achieved at the required level

Better than satisfactory (3,5) – the assumed learning outcomes have been achieved at the average required level

Satisfactory (3,0) – the assumed learning outcomes have been achieved at the minimum required level

Unsatisfactory (2,0) – the assumed learning outcomes have not been achieved

Course description

Part 2

Other useful information about the course		
12. Name of Department, mailing address, e-mail: Department of Molecular Biology and Genetics, ul. Medyków 18; 40-752 Katowice; biolmolgen@sum.edu.pl		
13. Name of the course coordinator: Dr Karolina bajdak-Rusinek		
14. Prerequisites for knowledge, skills and other competencies: Knowledge of basic concepts in the field of genetics at the high school level Knowledge of molecular genetic mechanisms		
15. Number of students in groups	In accordance with the Senate Resolution	
16. Study materials	Syllabus, handbooks, materials from assistant, materials in elearning platform https://elearning.sum.edu.pl/ ; “Genetics II US” course.	
17. Location of classes	Department of Molecular Biology and Genetics, bldg. C1; 4th floor; Seminar room 400 and Didactics and Medical Simulation Center (DMSC)	
18. Location and time for contact hours	Department of Molecular Biology and Genetics, bldg. C1; 4th floor; Tuesdays 13.00-14.00.	
19. Learning outcomes		
Number of the course learning outcome	Course learning outcomes	Reference to learning outcomes indicated in the standards
1.	Knows autosomal and heterosome aberrations that cause diseases, including oncogenesis and cancer	C.W7
2.	Knows the diagnosis of gene and chromosomal mutations responsible for hereditary and acquired diseases, including cancer;	C.W9
3.	Knows the indications for genetic tests carried out in order to individualize pharmacotherapy;	C.W41
4.	Knows the basic directions of therapy development, in particular possibilities of cell, gene and targeted therapy in specific diseases	C.W42
5.	Knows the causes, symptoms, principles of diagnosis and therapeutic procedure in the case of the most common genetic syndromes in children.	E.W3.10
6.	Knows the possibilities of modern cancer therapy including multimodal therapy, cell and gene therapy perspectives and their	E.W25

	undesirable effects	
7.	Knows the causes, symptoms, principles of diagnosis and therapeutic procedure in the most common hereditary diseases;	E.W37
8.	Is able to use data bases, including the internet databases and is able to search required information with accessible tools.	B.U10
9.	Make decisions about the need for cytogenetic and molecular tests	C.U3
10.	Estimate the risk of disclosure of the disease in the offspring based on family predisposition and the impact of environmental factors	C.U5
20. Forms and topics of classes		Number of hours
21.1. Lectures		20
1. SYNDROMES OF CHROMOSOMAL INSTABILITY		3
2. HEREDITARY CANCERS		3
3. EPIGENETICS		3
4. MULTIFACTORIAL DISEASES		3
5. RARE DISEASES		3
6. PRENATAL DIAGNOSIS		3
7. PRINCIPLES OF GENETIC COUNSELING		2
22.2. Seminars		20
1. Dysmorphology, part 1		3
2. Dysmorphology, part 2		3
3. Hereditary cancer syndromes		3
4. Genetic blood disorders		3
5. Gene therapy		3
6. Cancer biology and genetics		3
7. How to read pedigree		2
23.3. Labs		20
1. Genetic databases		3
2. Chromosomal organization of human genome. Principles of cytogenetic nomenclature		3
3. Numerical and structural aberrations of chromosomes. Cytogenetic techniques.		3
4. Gene mutations and diagnostic methods		3
5. Hereditary cancers		3
6. Pedigrees (Autosomal, sex-linked and mitochondrial Inheritance).		3
7. Final test		2
24. Readings		
1. Medical Genetics – Lynn B. Jorde, Johan C. Carey, Michael J. Bamshad		
2. Emery's Elements of Medical Genetics. R.F. Mueller, S.D. Young; Ed. Churchill Livingstone		
3. Materials in elearning platform https://elearning.sum.edu.pl/ ; "Genetics II US" course		
25. Detail evaluation criteria		
In accordance with the recommendations of the inspection bodies Completion of the course – student has achieved the assumed learning outcomes Detail criteria for completion and evaluation of the course are specified in the course regulations		