INFORMATION ABOUT THE COURSE

Genetics

1. Basic information

Field of studies field of medical and health sciences, discipline: medical sciences **Unit responsible for the field of studies** Faculty of Medicine Bydgoszcz University of Science and Technology **Level of studies** Uniform master's studies **Profile of studies** General academic **Form of studies** Full-time	**Studies cycle** …….. **Course code** 17-EMS-GEN-SP1 **Language** English **Obligatory** Yes

Prerequisites	Subject knowledge and skills: Biochemistry (Understanding the structure and properties of nucleic acids and other organic compounds is crucial to understanding the mechanisms of genetic inheritance) and Molecular Biology (Students should be familiar with the basic principles of molecular biology, such as DNA structure, gene organisation, and the processes of replication, transcription and translation). Method of verification: Passing the subjects defined as introductory is equivalent to fulfilling the prerequisites for the subject.
Introductory courses	Biochemistry, Molecular biology
Coordinator	Anna Kloska, PhD

Study period	**Form of assessment** **Form and hours of classes**	**ECTS credits**
Winter semester	Exam Lecture 45h Exercise 30h Seminar 20h	7.0

2. Learning outcomes

Code	**Description of learning outcomes**	**Learning outcomes reference**
Knowledge (student knows and understands):		
K1	development, structure and function of the human body in normal and pathological conditions	O.W1.
K2	symptoms and course of genetic diseases	O.W2.
K3	diagnostic and therapeutic modalities specific to genetic conditions	O.W3.
K4	the normal human karyotype and the different types of sex determination	C.W1.

K5	genetic causes of hereditary predisposition to cancer	C.W2.
K6	principles of inheritance of different numbers of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of non-nuclear genetic information	C.W3.
K7	Genetic determinants of human blood groups and serological conflict in the Rh system	C.W4.
K8	genetic determinants of the most common single-gene, polygenic and multifactorial diseases, major chromosome aberration syndromes, syndromes caused by genomic rearrangements, polymorphisms, epigenetic and post-transcriptional changes	C.W5.
K9	factors affecting the primary and secondary genetic balance of the population	C.W6.
K10	genetic determinants of congenital malformations and selected rare diseases and the possibility of their prevention	C.W7.
K11	genetic diagnostic methods and their basic indications	C.W8.
K12	Genetic mechanisms for the acquisition of drug resistance by microorganisms and cancer cells and their relationship to the need to individualise pharmacotherapy	C.W9.
K13	pathogenesis of diseases, including genetic and environmental conditions	C.W26.
K14	options and types of biological, cellular, gene and targeted therapies for specific diseases	C.W33.
K15	causes, symptoms, principles of diagnosis and therapeutic management of the most common genetic diseases in children and adults	E.W36.
K16	benefits and limitations of laboratory tests	E.W40.
Abilities (student can do/perform):		
A1	recognise medical problems and identify priorities for medical management	O.U1.
A2	plan the diagnostic procedure and interpret the results	O.U3.
A3	communicate with the patient and his/her family in an atmosphere of trust, taking into account the needs of the patient, and convey adverse information, using the principles of professional communication	O.U7.
A4	chart and analyse pedigrees and identify clinical and pedigree features suggestive of a genetic basis for diseases	C.U1.
A5	decide on the need for cytogenetic and molecular tests	C.U2.
A6	read basic genetic test results, including karyotypes	C.U3.

A7	determine genetic risk based on pedigree and genetic test result for chromosome aberrations, genomic rearrangements, single-gene and multifactorial diseases	C.U4.
Social skills (the student is ready to):		
S1	is willing to be guided by the welfare of the patient	O.K2.
S2	is willing to respect medical confidentiality and patients' rights	O.K3.
S3	is ready to act towards the patient on the basis of ethical principles, with an awareness of the social determinants and limitations of the disease	O.K4.
S4	is willing to draw conclusions from his own measurements or observations	O.K8.

3. Programme contents

No.	Programme contents	Form of studies	Learning outcomes covered by the programme content
1	Introduction to genetics: basic concepts and definitions	Lecture	K1, K2, K3, K4
2	Development, structure and function of the human body in normal and pathological conditions	Lecture	K1, K2, K3
3	Symptoms and course of genetic diseases Human karyotype and types of sex determination	Lecture, Exercise	K2, K3, K4, K5, K13, A1, A2, A5, A6, S1
4	Genetic causes of hereditary predisposition to cancer	Lecture, Seminar	K1, K2, K3, K5, K6, K8, K9, K11, K13, A1, A2, A3
5	Principles of inheritance of traits, including single-gene and polygenic inheritance Genetic determinants of the most common monogenic and polygenic diseases	Lecture, Seminar, Exercise	K1, K2, K3, K5, K6, K8, K9, K11, K13, K15, A1, A2, A3, A5, S1, S2, S3, S4
6	Genetic determinants of human blood groups and serological conflict in the Rh system	Lecture, Seminar, Exercise	K2, K3, K7, K16, A1, A2, S1, S2, S3, S4
7	Genetic determinants of malformations and rare diseases	Lecture, Seminar	K1, K2, K3, K5, K6, K8, K16, A1, A2, A7

8	Methods of genetic diagnosis Presentation of new methods of genetic diagnosis and therapy	Lecture, Exercise	K1, K2, K3, K11, K15, A1, A2, A5, A6, A7, S1, S2, S3, S4
9	Genetic mechanisms of drug resistance	Lecture, Seminar	K2, K3, K11, K12, K14, A1, A2, S1, S2, S3, S4
10	Pathogenesis of diseases: genetic and environmental determinants	Lecture, Seminar	K1, K2, K3, K5, K8, K11, K13, K15, A1, A2, A7, S1, S2, S3, S4
11	Biological, cellular, gene and targeted therapies for genetic diseases	Lecture, Seminar, Exercise	K1, K2, K3, K5, K6, K8, K11, K12, K16, A1, A2, A3, A5, A6, A7, S1, S2, S3, S4
12	Fetal diagnosis. Laboratory exercises with genetic diagnostic techniques Discussion of controversial aspects in genetic diagnosis	Lecture, Seminar, Exercise	K1, K2, K3, K5, K6, K8, K11, K12, K16, A1, A2, A3, A5, A6, A7, S1, S2, S3, S4
13	Genetically determined diseases in children and adults: causes, symptoms, diagnosis and therapy	Lecture, Seminar, Exercise	K1, K2, K3, K5, K6, K9, K10, K11, K12, K16, A1, A2, A3, A6, A7, S1, S2, S3, S4
14	Possibilities and limitations of laboratory tests in genetics Medical problems and medical management in genetics Planning the diagnostic procedure Decisions regarding genetic testing. Interpretation of genetic test results, including karyotypes. Determination of genetic risk Discussion of genetic test results and their implications for patients	Lecture, Seminar, Exercise	K1, K2, K3, K8, K9, K10, K11, K13, K15, A1, A2, A3, A5, A7, S1, S2, S3, S4
15	Communication with the patient and family in the context of genetic diseases. Conversation with patients regarding diagnosis and treatment of genetic diseases Maintenance of medical records	Seminar, Exercise	K1, K2, K3, K15, K16, A1, A2, A3, S1, S2, S3, S4
16	Analysis of pedigrees and clinical characteristics Plotting and analysis of patients' pedigrees Genetic risk assessment based on pedigree and test results Investigation of cases of familial burden of genetic diseases	Lecture, Seminar, Exercise	K2, K3, K8, K9, K11, K12, K13, K15, A1, A2, A3, A4, A5, A6, A7, S1, S2, S3, S4

4. Methods of verifying and assessing the learning outcomes achieved by the student

Winter semester

Form of studies		
Lecture	**Methods of studies form:**	
	Lecture	
	Methods of verification:	**Involvement:**
	Written exam	100%
	Conditions for passing the course:	
	A prerequisite for passing the course is a positive grade from a written examination. Written examination in the form of a test (50 single-choice questions, pass mark 60%). A prerequisite for passing the exam is passing the exercises and seminars.	
Exercise	**Methods of studies form:**	
	Laboratory exercise, Discussion	
	Methods of verification:	**Involvement:**
	Colloquium	90%
	Activity	5%
	Observation	5%
	Conditions for passing the course:	
	Colloquium in the form of a test (50 single-choice questions, pass mark 60%). Attendance and activity in classes is a prerequisite for passing the test.	
Seminar	**Methods of studies form:**	
	Discussion, Demonstration, Case study, Group work	
	Methods of verification:	**Involvement:**
	Presentation	60%
	Activity	20%
	Observation	20%
	Conditions for passing the course:	
	A prerequisite for successful completion of the seminars is the preparation of a presentation on a topic assigned by the instructor. Student activity is assessed in the seminars.	

Learning outcomes	Methods of verification

	Written exam	Activity	Colloquium	Observation	Presentation
K1	X		X		X
K2	X		X		X
K3	X		X		X
K4	X		X		X
K5	X		X		X
K6	X		X		X
K7	X		X		X
K8	X		X		X
K9	X		X		X
K10	X		X		X
K11	X		X		X
K12	X		X		X
K13	X		X		X
A1		X	X	X	X
A2		X	X	X	
A3		X		X	
A4		X		X	
A5		X	X	X	
A6		X		X	
A7		X		X	
A8		X		X	
A9		X		X	
A10		X		X	
S1		X	X	X	X
S2		X		X	
S3		X		X	
S4		X	X	X	

5. Student workload – balance of hours and ECTS credits

Students activity		**Student workload Number of hours**
	Lecture	45

Classes conducted with the direct participation of an academic teacher or other persons conducting classes	Exercise	30
	Seminar	20
Student's own work	Preparing for classes	15
	Preparing a presentation	10
	Studying literature	15
	Preparing for a test	20
	Preparing for an exam	20
Total student workload		175
ECTS		7

One (teaching) hour is 45 minutes.

6. Literature

The list of required and recommended literature will be provided by the lecturer at the first meeting.