



# Pomeranian Medical University in Szczecin

## SYLLABUS of the MODULE (SUBJECT)

### General Information

Code		Title	<i>Clinical Genetics</i>
Module type			<i>Obligatory</i>
Faculty			<i>Faculty of Medicine</i>
Field of study			<i>medicine</i>
Major			<i>Not applicable</i>
Level of study			<i>II level/long-cycle (S2J)</i>
Mode of study			<i>intramural</i>
Year of study			<i>IV</i>
Semester			<i>block</i>
ECTS points			<i>5</i>
Types of training			<i>seminars/classes 65 h (35h seminars; 30 h classes)</i>
Tutor responsible for the module			<i>Prof. dr hab. n. med. Jan Lubiński</i>
Tutors conducting the subject			Prof. dr hab. n. med. Jan Lubiński Prof. dr hab. n. med. Cezary Cybulski (cezarycy@sci.pum.edu.pl) Prof. dr hab. n. med. Jacek Gronwald, (jgron@sci.pum.edu.pl) Prof. dr hab. n. med. Tadeusz Dębniak, (debniak@sci.pum.edu.pl) dr n. med. Urszula Teodorczyk (urteo@pum.edu.pl) dr n. med. Katarzyna Gołębowska (k.tutlewska@wp.pl) mgr Tomasz Gromowski (tomaszbiotech@gmail.com) dr n. med. Aniruddh Kashyap (kashyap.aniruddh@gmail.com) mgr Katarzyna Kaczmarek (katarzyna.kaczm@gmail.com) dr n. med. Wojciech Kluźniak (dante.k@wp.pl) dr n. med. Helena Rudnicka (helena.rudnicka@pum.edu.pl) lek. med. Krzysztof Bernatowicz (ka.bernatowicz@gmail.com) dr Krzysztof Kąklewski (chris2k@wp.pl) Nina Peruga (nina.perugina@gmail.com) Aleksandra Pietrzyk (olapietrzyk@wp.pl) Karolina Prajzencanc (karolina.praj@wp.pl) Bogna Rusak (b_rusak@yahoo.com) Elżbieta Złowocka-Perłowska (elzunik@wp.pl)
WWW			www.genetyka.com
Language			English

## Detailed information

Module objectives		It is to provide the basic knowledge in the field of modern clinical genetics with respect to any branch of medicine. Knowledge should include the rules of inheritance and diagnosis of hereditary diseases, the identification of genes responsible for the formation of genetic diseases, mechanisms of regulation of gene expression; mechanisms of DNA repair.
Prerequisite /essential requirements/	Knowledge	Knowledge of the: basic concepts of genetics, chromosome structure and description of proper human karyotype .
	Skills	To be able to solve problems with use of the Mendel laws. Knowledge of the basic concepts of genetics, chromosome structure and description of proper human karyotype
	Competences	The habit of self-study, group work

Description of the learning outcomes for the subject /module			
Number of learning outcome	Student, who has passed the (subject) Knows /is able to /can:	SYMBOL (referring the standards) EKK	Method of verification of learning outcomes
KL2JPW01	knows the basic concepts of genetics	K_C.W1	Multiple choice test. Exam
KL2JPW02	describes phenomena of coupling and cooperation of genes	K_C.W2	Multiple choice test. Exam
KL2JPW03	correctly describes human karyotype and different stages of sex determination	K_C.W3	Multiple choice test. Exam
KL2JPW04	describes structure of chromosomes and molecular base of mutagenesis	K_C.W4	Multiple choice test. Exam
KL2JPW05	knows the principles of inheritance of a variety of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of extra-nuclear genetic information	K_C.W5	Multiple choice test. Exam
KL2JPW06	knows genetic conditions of human blood groups and Rh complex serological incompatibility	K_C.W6	Multiple choice test. Exam
KL2JPW07	describes aberration of autosomes and heterosomes causing diseases, incl. oncogenesis	K_C.W7	Multiple choice test. Exam
KL2JPW08	knows factors affecting primary and secondary genetic equilibrium of population	K_C.W8	Multiple choice test. Exam
KL2JPW09	knows principles for diagnostics of gene and chromosome mutations responsible for hereditary diseases, incl. neoplasms	K_C.W9	Multiple choice test. Exam
KL2JPW10	defines advantages and threats related to GMO's found in ecosystem	K_C.W10	Multiple choice test. Exam
KL2JPW11	knows genetic mechanism for contracting drug-resistance by micro-organisms and malignant cells	K_C.W11	Multiple choice test. Exam
KL2JPW12	defines clinical course of specific and non-	K_C.W27	Multiple choice test.

	specific inflammation and describes the processes of regeneration of tissues and organs		Exam
KL2JPW13	knows indications for genetic examination in order to individualize pharmacotherapy	K_C.W40	Multiple choice test. Exam
KL2JPW14	knows key directions of therapy development, in particular the potential of cellular therapy, gene therapy and target therapy in certain diseases	K_C.W41	Multiple choice test. Exam
KL2JPW15	knows the basic principles of ethics in genetics		Multiple choice test. Exam
KL2JPW16	has knowledge of fetal development and developmental defects		Multiple choice test. Exam
KL2JPW17	He knows the symbols of pedigree description		Multiple choice test. Exam
KL2JPU01	analyses genetic crosses and genealogy of human traits and diseases and evaluates risk of the birth of chromosome aberration affected children	K_C.U1	Multiple choice test. Exam
KL2JPU02	identifies indications during prenatal examination	K_C. U2	Multiple choice test. Exam
KL2JPU03	makes decision on cytological and molecular examination	K_C.U3	Multiple choice test. Exam
KL2JPU04	performs morphometric measurements, analyzes morphograms and notes karyotypes of diseases	K_C.U4	Multiple choice test. Exam
KL2JPU05	assesses risk of the occurrence of disease in progeny on the basis of family predispositions and influence of environmental factors	K_C.U5	Multiple choice test. Exam
KL2JPU06	Uses a code of ethical conduct in the field of genetics		Multiple choice test. Exam
KL2JPU07	Can read the correct DNA bases sequence		Multiple choice test. Exam
KL2JPU08	Knows how to conduct interview with the patient		Multiple choice test. Exam
KL2JPU09	can make and analyze pedigree on the basis of interview with the patient		Credit
KL2JPK01	accepts the need for standards of conduct	K_K01	Credit
KL2JPK02	recognizes concept and need for responsibility for property he/she has been entrusted with	K_K02	Credit
KL2JPK03	Demonstrates the awareness for self-education, understands the need for continuing professional education, can inspire and organize learning processes in others	K_K03	Credit
KL2JPK04	co-operates with team members; can co-operate within a group and take different roles	K_K04	Credit
KL2JPK05	adheres to proper examiner/examinee relationship while performing functional tests and observations	K_K05	Credit

**Matrix presenting the learning outcomes of the subject/module in relation to the form of classes**

Number of learning outcome	Student, who has passed the (subject) Knows /is able to /can:	Types of training							
		Lecture	Seminar	Laboratory classes	Project work	Clinical classes	Classes	Practical classes	Other
KL2JPW01	knows the basic concepts of genetics		X			X			
KL2JPW02	describes phenomena of coupling and cooperation of genes		X			X			
KL2JPW03	correctly describes human karyotype and different stages of sex determination		X			X			
KL2JPW04	describes structure of chromosomes and molecular base of mutagenesis		X			X			
KL2JPW05	knows the principles of inheritance of a variety of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of extra-nuclear genetic information		X			X			
KL2JPW06	knows genetic conditions of human blood groups and Rh complex serological incompatibility		X			X			
KL2JPW07	describes aberration of autosomes and heterosomes causing diseases, incl. oncogenesis		X			X			
KL2JPW08	knows factors affecting primary and secondary genetic equilibrium of population		X			X			
KL2JPW09	knows principles for diagnostics of gene and chromosome mutations responsible for hereditary diseases, incl. neoplasms		X			X			
KL2JPW10	defines advantages and threats related to GMO's found in ecosystem		X			X			
KL2JPW11	knows genetic mechanism for contracting drug-resistance by micro-organisms and malignant cells		X			X			
KL2JPW12	defines clinical course of specific and non-specific inflammation and describes the processes of regeneration of tissues and organs		X			X			
KL2JPW13	knows indications for genetic examination in order to individualize pharmacotherapy		X			X			
KL2JPW14	knows key directions of therapy development, in particular the potential of cellular therapy, gene therapy and target therapy in certain diseases		X			X			
KL2JPW15	knows the basic principles of ethics in genetics		X			X			
KL2JPW16	has knowledge of fetal development and developmental defects		X			X			
KL2JPW17	He knows the symbols of pedigree description		X			X			
KL2JPU01	analyses genetic crosses and genealogy of human traits and		X			X			

	diseases and evaluates risk of the birth of chromosome aberration affected children								
KL2JPU02	identifies indications during prenatal examination		X			X			
KL2JPU03	makes decision on cytological and molecular examination		X			X			
KL2JPU04	performs morphometric measurements, analyzes morphograms and notes karyotypes of diseases		X			X			
KL2JPU05	assesses risk of the occurrence of disease in progeny on the basis of family predispositions and influence of environmental factors		X			X			
KL2JPU06	Uses a code of ethical conduct in the field of genetics		X			X			
KL2JPU07	Can read the correct DNA bases sequence		X			X			
KL2JPU08	Knows how to conduct interview with the patient		X			X			
KL2JPU09	can make and analyze pedigree on the basis of interview with the patient		X			X			
KL2JPK01	accepts the need for standards of conduct		X			X			
KL2JPK02	recognizes concept and need for responsibility for property he/she has been entrusted with		X			X			
KL2JPK03	Demonstrates the awareness for self-education, understands the need for continuing professional education, can inspire and organize learning processes in others		X			X			
KL2JPK04	co-operates with team members; can co-operate within a group and take different roles		X			X			
KL2JPK05	adheres to proper examiner/examinee relationship while performing functional tests and observations		X			X			

Module (subject) contents		
Symbol of teaching programme	Content of teaching programme	References to learning outcomes
TK01	Indications for genetic testing. Principles of cytogenetic studies. Chromosome construction, karyotype, chromosome aberrations	KL2JPW01; KL2JPW02; KL2JPW03; KL2JPW04; KL2JPW05; KL2JPW06; KL2JPW07; KL2JPW08; KL2JPU01; KL2JPU02; KL2JPU03; KL2JPU04; KL2JPU05; KL2JPU06; KL2JPU07; KL2JPU08; KL2JPU09; KL2JPK05
TK02	Genetics of disorders of sex differentiation and determination and failures in reproduction.	KL2JPW03; KL2JPW04; KL2JPW05; KL2JPK01
TK03	Molecular techniques for detecting mutations	KL2JPU01; KL2JPW04; KL2JPW05; KL2JPU06; KL2JPU07; KL2JPU08
TK04	Hereditary cancers. Principles of inheritance and diagnosis of hereditary diseases; Mitochondrial diseases	KL2JPW05; KL2JPW07; KL2JPU08; KL2JPU09; KL2JPW14; KL2JPW17; KL2JPK02
TK05	Pedigree clinical criteria, indications for DNA tests.	KL2JPW05; KL2JPW09; KL2JPW13; KL2JPU09; KL2JPU08; KL2JPU07; KL2JPK05

TK06	Hereditary cancer: - breasts - colon -prostate Von Hippel-Lindau syndrom - neurofibromatosis type NF1, NF2	KL2JPW05; KL2JPW09; KL2JPW11; KL2JPW 12; KL2JPW13; KL2JPW14; KL2JPU01; KL2JPU09; KL2JPK02; KL2JPK04
TK07	Von Willebrand syndrom. Homologous recombination and gene therapy	KL2JPW09; KL2JPW14; KL2JPU09; KL2JPU08; KL2JPU07; KL2JPU06; KL2JPK02; KL2JPK04
TK08	Monogenic diseases -autosomal recessive. -autosomal dominant. Chromosomal X-linked monogenic diseases. Multigene diseases. Chromosomal diseases.	KL2JPW02; KL2JPW05; KL2JPW07; KL2JPW09; KL2JPW14; KL2JPU01; KL2JPU08; KL2JPU09; KL2JPK02
TK09	Malformations. Prenatal Diagnosis. Causes. Concepts.	KL2JPW09; KL2JPW13; KL2JPW16; KL2JPW17; KL2JPU07; KL2JPU08; KL2JPU09; KL2JPK04
TK10	Ethics in genetics, legal aspects of genetic examinations.	KL2JPW15; KL2JPU06; KL2JPK03

#### References and educational resources

1. Molecular Cell Biology/Harvey Lodish W.H.Freeman;7 ED. cop. 2012 r ISBN 9781429234139

2. Essential medical genetics/ Edward S. Tobias, Michael Connor, Malcolm Ferguson-Smith. 6th ed Chichester;Wiley-Blackwell.cop.2011 ISBN 9781405169745

3. . Medical genetics / Lynn B. Jorde, John C. Carey, Michael J. Bamshad. 5th ed. Philadelphia : Elsevier, cop. 2016; ISBN 978-0-323-18835-7

4. Genetics in Medicine/Thompson & Thompson. Robert L.Nussbaum, Roderick R. McInnes, Huntington F.Willard. 8th ed. Philadelphia : Elsevier, cop. 2016; ISBN 978-1-4377-0696-3

#### Student's workload (balance sheet of ECTS points)

Form of student's activity (in-class participation; activeness, produce a report, etc.)	Workload [h]		
	Tutor	Student	Average
activities that require direct participation of tutors		65	
Preparation to the classes			
Reading of the indicated/specified literature			
Report writing/project making			
Time spent to prepare for the exam			
Other			
Student's workload in total			
<b>ECTS points for the subject</b>	<b>5</b>		

#### Remarks at the end

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Methods of assessment, for example:

E – exam- problem resolving

S – verifying of practical skills

R – report

D – discussion

P – presentation

Others-