Appendix to Decision No. 4/2020

Pomeranian Medical *University* in *Szczecin[PUM]*

COURSE SYLLABUS

General information

Name of Course: Clinical genetics	
Type of activity	Compulsory
PUM Faculty	Faculty of Medicine and Dentistry
Field of study	Medicine
Specialization	-
Level of study	Long-term studies
Form of study	full-time
Year of study /semester	4/ winter and summer semester (BLOCK)
Number of ECTS credits allocated	4,5
Forms of teaching (number of hours)	lectures (e-learning) - 10h seminars - 25h practical exercises - 30h
Methods of verification and assessment of learning outcomes *	☐graded credit: ☐descriptive credit ☐test credit ☐practical credit ☐oral credit ☐credit without grade ☒final exam: ☐descriptive credit ☒test credit ☐practical credit ☐oral credit
Head of Unit	Prof. dr hab. Jan Lubiński, Prof. PhD
Teaching assistant professor or person responsible for the course	Urszula Teodorczyk, PhD Elżbieta Kowalska, PhD
Name and contact details of unit	Department of Genetics and Pathomorphology ul. Unii Lubelskiej 171-252 Szczecin tel. 91 441-72-50 e-mail: lubinski@pum.edu.pl
Unit's website	https://www.pum.edu.pl/wydzialy/wydzial-lekarsko-biotechnologiczny/zaklad-genetyki-i-patomorfologii
Language of instruction	Polish/English

*tick as appropriate, changing ☐ to ☒.

Detailed information

Course objectives		The objective of the module is to provide basic knowledge of modern clinical genetics relevant to any branch of medicine. Among other things, it is important to know the principles of inheritance and diagnosis of hereditary diseases, identification of genes responsible for the formation of genetically determined diseases, mechanisms of gene regulation and expression; mechanisms of DNA damage repair, gene therapy, etc.
Preliminary requirements in terms of	Knowledge	Knowledge of basic concepts of genetics, chromosome structure and description of the normal human karyotype and analysis of pedigrees.
	Skills	Ability to solve genetic crosses and pedigrees of human traits and diseases and assess the risk of a child being born with chromosomal aberrations.
	Social competences	The habit of self-education, teamwork

LEARNING OUTCOMES			
Number of learning outcome	**A student who has completed of the COURSE knows/can:**	**SYMBOL (reference to) learning outcomes for the field of study**	**Method of verifying the learning outcomes***
W01	A student knows the basic concepts of genetics	K_C.W1	ET
W02	A student describes the phenomena of gene coupling and interaction	K_C.W2	ET
W03	A student describes the normal human karyotype and the different types of sex determination	K_C.W3	ET
W04	A student describes the structure of chromosomes and the molecular basis of	K_C.W4	ET
W05	knows the principles of inheritance of different numbers of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of extra-nuclear genetic information	K_C.W5	ET
W06	A student knows genetic conditions of human blood groups and serological conflict in Rh system	K_C.W6	ET
W07	A student describes the aberrations of autosomes and heterosomes that cause disease, including cancer oncogenesis	K_C.W7	ET

W08	A student knows the factors influencing the primary and secondary genetic balance of populations	K_C.W8	ET
W09	A student knows the basis of diagnosis of gene and chromosome mutations responsible for inherited and acquired diseases, including cancer	K_C.W9	ET
W10	A student knows the indications for genetic testing in order to individualise	K_C.W40	ET
W11	A student knows the basic directions of therapy development, in particular the possibilities of cell, gene and targeted therapy in specific diseases	K_C.W41	ET
W12	A student knows the basic principles of ethics in the field of genetics		ET
W13	A student has knowledge of foetal development and malformations		ET
W14	A student knows the symbols for the description of the pedigrees		ET
U01	A student analyses genetic crosses and pedigrees of human traits and diseases and assesses the risk of a child being born with chromosome aberrations	K_C.U1	ET
U02	A student identifies indications for prenatal tests	K_C.U2	ET
U03	A student decides on the need for cytogenetic and molecular tests	K_C.U3	ET, O
U04	A student performs morphometric measurements, analyses the morphogram and records disease karyotypes	K_C.U4	ET , O
U05	A student estimates the risk of a given disease in offspring based on family predisposition and the influence of environmental factors	K_C.U5	ET
U06	A student applies the code of ethical conduct in the field of genetics		ET
U07	A student is able to interview the patient		O
U08	A student knows how to prepare and analyse pedigrees based on patient interview		ET
K01	A student accepts the need for ethical standards;	K_K01	O
K02	A student understands the concept and the need for responsibility for the entrusted	K_K02	O
K03	A student shows the habit of self-education, understands the need of lifelong learning, is able to inspire and organize the learning process of others	K_K03	O

K04	A student cooperates with team members; can cooperate in a group taking up different roles	K_K04	O
K05	A student ,during observations and functional tests, observes proper relations between the examiner and the examined	K_K05	O

Table of learning outcomes in relation to the form of classes

Number of learning outcome	Learning outcomes	Form of the classes						
		Lecture (e-l)	Seminar	Practical	Clinical exercises	Simulations	E-learning	Other forms
W01	K_C.W1		X		X			
W02	K_C.W2		X		X			
W03	K_C.W3		X					
W04	K_C.W4		X					
W05	K_C.W5		X					
W06	K_C.W6		X					
W07	K_C.W7		X					
W08	K_C.W8		X					
W09	K_C.W9		X					
W10	K_C.W40		X					
W11	K_C.W41		X					
W12			X					
W13			X					
W14			X					
U01	K_C.U.1		X		X			
U02	K_C.U2		X					
U03	K_C.U3		X		X			
U04	K_C.U4		X		X			
U05	K_C.U5		X					
U06			X					
U07			X		X			
U08			X					
K01	K_K01		X		X			
K02	K_K02		X		X			

K03	K_K03		X		X			
K04	K_K04				X			
K05	K_K05		X		X			

TABLE OF CURRICULUM			
Curriculum number	**Curriculum content**	**Number of hours**	**Reference to the learning outcomes for the CLASSES**
Winter and summer semester (BLOCK)			
Lectures - 10h			
TK01	Indications for genetic testing	1h	W01, W02, W03, W04, W05, W06, W07, W08
TK02	Genetics of disorders of sex differentiation determination and reproductive failure.	2h	W03, W04, W05, K03
TK03	Hereditary cancers.	1h	W05, W07, W11, W14, K03
TK04	Pedigree and clinical criteria, indications for DNA tests	1h	W05, W09, W10, W14, K03
TK05	Hereditary cancer: - Neurofibromatosis types NF1, NF2	1h	W02, W05, W07, W09, W10
TK06	von Willebrand's syndrome.	1h	W09, W10
TK07	Monogenic diseases - autosomal recessive. Autosomal-dominant diseases. Single-gene diseases coupled to the X chromosome. Multigene diseases. Chromosomal diseases.	2h	W02, W05, W07, W09, W11
TK08	Developmental defects. Causes. Concepts.	1h	W09, W10, W13
Seminars - 25h			
TK01	Principles of cytogenetic research. Chromosome structure, karyotype, chromosome aberrations.	3h	W01, W02, W03, W04, W05, W06, W07, W08, U01, U02, U03, U04, U05, K03

TK02	Genetics of disorders of sex differentiation determination and reproductive failure.	2h	W03, W04, W05, K01, K03
TK03	Principles of inheritance and diagnosis of inherited diseases; mitochondrial diseases	3h	W05, W07, W11, W14, K02, K03
TK04	Pedigree and clinical criteria, indications for DNA tests	3h	W05, W09, W10, W14, U07, U08, K03
TK05	Hereditary cancer: - breasts - colon - prostates - von Hipell-Lindau syndrome - Neurofibromatosis types NF1, NF2	3h	W02, W05, W07, W09, W10, U01, U05
TK06	Homologous recombination and gene therapy	2h	W09, W10, U06
TK07	Monogenic diseases - autosomal recessive. Autosomal-dominant diseases. Single-gene diseases coupled to the X chromosome. Multigene diseases. Chromosomal diseases.	4	W02, W05, W07, W09, W11, U01, U08, K02
TK08	Malformations. Prenatal diagnosis. Causes. Concepts.	4	W09, W10, W13, U02, U03, U04, U05
TK09	Ethics in genetics	1h	W12, U06, U07, K01, K02, K05
Practical Exercises - 30h			
TK01	Exercises in Oncology Genetic Outpatient Clinic	30	W01, W02, W05, W07, W12, W14, U06, U07, U08, K01, K02,K03, K04, K05

Recommended Literature:
Required literature:
1. „Podstawy Genetyki Medycznej” S. H. Connor; M. A. Ferguson
2. „Genetyka kliniczna nowotworów 2017, 2018” monografia (red.) J. Lubiński
3. "Medical genetics" LynnB.Jorde, John C. Carey, Michael J. Bamshad , Wroclaw 2014 Edra Urban & Partner.
4. „Genetyka medyczna”, redakcja naukowa wyd. pol. Anna Latos-Bieleńska, PZWL 2013

Supplementary literature:
1. „Genetyka” J. M. Friedman; F. J. Gill i inni (red.) J. Limona
2. Genetyka człowieka. Rozwiązanie problemów medycznych” B. R. Korf PWN Warszawa 2003.
3. „Biologia molekularna w medycynie, elementy genetyki klinicznej” red. Nauk. Jerzy Bal, PWN, Warszawa 2011.

Student workload	
Form of student workload (class participation, activity, report preparation, etc.)	Student workload [h]. In the teacher's assessment (opinion)
Contact hours with the teacher/instructor	65
Preparation for exercise/seminar	2
Reading the indicated literature	8
Writing a lab/exercise report/preparing a project/reference, etc.	
Preparation for a test/short test	5
Preparing for the exam	50
Other	
Total student workload	130
ECTS credits	4,5
Notes	

*Example Methodss of verification of educational outcomes:
EP - written exam
EU - oral test
ET - test exam
EPR - practical test
K - test
R - paper
S - testing of practical skills
RZĆ - report from practical exercises with discussion of results
O - evaluation of student's activity and attitude
SL - Laboratory report
SP - case study
PS - assessment of ability to work independently
W - short test before the beginning of classes
PM - multimedia presentation
and other