SYLLABUS – A COURSE DESCRIPTION

I. General informaion

1. Course name: **Human diseases – epidemiology, etiology and therapy**
2. Course code: **01-BTA-HUMANDIS**
3. Course type (compulsory or optional): **optional**
4. Study programme name: **Biotechnology**
5. Cycle of studies (1st or 2nd cycle of studies or full master's programme): **2nd cycle of studies**
6. Educational profile (general academic profile or practical profile): **general academic profile**
7. Year of studies (if relevant): **II**
8. Type of classes and number of contact hours (e.g. lectures: 15 hours; practical classes: 30 hours):
   lectures: 15 hours
   practical classes: 15 hours
9. Number of ECTS credits: **3**
10. Name, surname, academic degree/title of the course lecturer/other teaching staff:
    dr hab. Julia Durzyńska, juliadur@amu.edu.pl
    dr hab. Małgorzata Borowiak, malgorzata.borowiak@amu.edu.pl
    dr hab. Mirosława Siatecka, siatecka@amu.edu.pl
11. Language of classes: **English**
12. Online learning – yes (partly – online / fully – online) / no:

II. Detailed information

1. Course aim (aims)

The principle goals of learning are:

1. To familiarize students with medical history of cancer, and classification of cancer; to familiarize students with classification of rare diseases (nuclear and mitochondrial); to familiarize students with the concept of rare diseases and metabolic syndromes: definitions, frequency, examples, components, risk factors and causes; to familiarize students with pre-diabetes and diabetes: classification, prevalence, etiology, and molecular mechanisms.

2. To explain actions of mutagens (chemical, physical, viral), DNA adducts, types of DNA damage, types of mutations in cancer; to explain mechanisms of DNA damage repair and related diseases when these mechanisms are defective; to explain genetic basis of rare metabolic diseases: de novo mutations and hereditary mutations. Chromosomes - types, division into groups, karyotypes and idiograms; to familiarize students with genetic instability, examples of chromosomal translocation and hybrid genes in several types of cancer; to familiarize students with chromosome structure abnormalities diseases: e.g. microdeletion, microduplication syndromes; to explain chromosome number abnormalities: trisomies, monosomies, chromosome instability, syndromes such as Bloom Syndrome, Fanconi Anemia, Nijmegen Syndrome - etiology and diagnostics.

3. To explain cell cycle control by products of proto-oncogenes and tumor suppressor genes; to familiarize students with oncogenes – how their activity is altered in cancer cells.

4. To familiarize students with colon cancer as a multistage process involving oncogenes and tumor suppressor genes; cancer-susceptibility genes in APC: concepts of gatekeepers, caretakers, landscapers; to explain the two-hits hypothesis of Knudson, statistical/mathematical deduction using an example of retinoblastoma, germ-line and somatic mutations; to explain inherited metabolic disorders; to explain genetic metabolic diseases: Gaucher, Fabry, Mucopolysaccharidosis, Pompe; to familiarize students with dynamic mutation diseases - neurodegenerative and neuromuscular.

5. To explain beta cell fault, insulin resistance, immune defect, insulin excels in developing diabetes; to familiarize students with regenerative medicine application for metabolic disorders; to explain atypical cases of metabolic diseases modeling in vitro;

to familiarize students with human mini-organs, organ-on-chip, multi-organ systems.

6. To familiarize students with epidemiology and diagnostics of cancer worldwide, rare and metabolic diseases; to explain diagnostic methods for sporadic and hereditary mutations causing human rare diseases.

7. To explain different cancer treatments: surgery, radiotherapy, chemotherapy, modern therapies (specific drugs blocking products of cancer genes, immunotherapies, oncolytic viruses); To explain personalized therapeutic nutrition for metabolic disorders.

2. Pre-requisites in terms of knowledge, skills and social competences (if relevant)

Before students start classes of human diseases – epidemiology, etiology and therapy they should know basics of: 1. human cell biology - cell cycle 2. biochemistry: structure of DNA and RNA, lipids, sugars 3. biochemical cycles and major metabolic cellular pathways 4. human gene structure and expression 5. cell signaling principles 6. principles of endocrinology and physiology

3. Course learning outcomes (EU) in terms of knowledge, skills and social competences and their reference to study programme learning outcomes (EK)

Course learning outcome symbol (EU)	On successful completion of this course, a student will be able to:	Reference to study programme learning outcomes (EK)
EU_01	use with understanding scientific terminology in the field of human carcinogenesis, metabolic and rare diseases.	BT_W04, BT_W09, BT_U05, BT_U04, BT_K01
EU_02	explain genetic background and etiology of human cancer, metabolic and rare diseases.	BT_W03, BT_U05, BT_U03, BT_K03
EU_03	knows epidemiology of human cancer, metabolic and rare diseases worldwide and can explain ethnic as well as socio-economic and other differences.	BT_W04, BT_W09, BT_U03, BT_U05, BT_K02, BT_K03
EU_04	explain different therapeutic approaches to treat human pathological conditions including cancer, metabolic and rare diseases; knows advanced models of human diseases.	BT_W01, BT_W06, BT_U01, BT_U03, BT_U05, BT_K01, BT_K02
EU_05	explanins how to: 1) monitor glucose uptake by different cell types in vitro, 2) perform karyotyping, chromosome identification, G-band staining and analyze relationships within a kindred. Pedigrees.	BT_W01, BT_U01, BT_U02, BT_U06, BT_K01, BT_K02
EU_06	find adequate scientific literature covering topics of human diseases – epidemiology, etiology and therapy and can express critical assessment.	BT_W09, BT_U02, BT_U04, BT_U05, BT_K01, BT_K02
EU_07	Select and use molecular techniques for diagnosis of human cancer, metabolic and rare diseases.	BT_W01, BT_W05, BT_U01, BT_U02, BT_K01

4. Learning content with reference to course learning outcomes (EU)

Course learning content	Course learning outcome symbol (EU)
medical history of cancer, and classification of cancer; classification of rare diseases (nuclear and mitochondrial); the concept of rare diseases: definitions, frequency; metabolic syndromes: definition, examples, components, risk factors and causes; pre-diabetes and	EU_01, EU_02, EU_06

diabetes: classification, prevalence, etiology, and molecular mechanisms.	
actions of mutagens (chemical, physical, viral), DNA adducts, types of DNA damage, types of mutations in cancer; mechanisms of DNA damage repair and related diseases when these mechanisms are defective; genetic basis of rare metabolic diseases: de novo mutations and hereditary mutations. Chromosomes - types, division into groups, karyotypes and idiograms; genetic instability, examples of chromosomal translocation and hybrid genes in several types of cancer; chromosome structure abnormalities diseases: e.g. microdeletion, microduplication syndromes; chromosome number abnormalities: trisomies, monosomies, chromosome instability syndromes such as Bloom Syndrome, Fanconi Anemia, Nijmegen Syndrome - etiology and diagnostics.	EU_02, EU_06, EU_05, EU_07
cell cycle control by products of proto-oncogenes and tumor suppressor genes; oncogenes – how their activity is altered in cancer cells.	EU_01, EU_02, EU_06, EU_03
colon cancer as a multistage process involving oncogenes and tumor suppressor genes; cancer-susceptibility genes in APC: concepts of gatekeepers, caretakers, landscapers; two-hits hypothesis of Knudson, statistical/mathematical deduction using an example of retinoblastoma, germ-line and somatic mutations; inherited metabolic disorders; to explain genetic metabolic diseases: Gaucher, Fabry, Mucopolysaccharidosis, Pompe; dynamic mutation diseases - neurodegenerative and neuromuscular - etiology and diagnostics.	EU_01, EU_02, EU_06, EU_07
beta cell fault, insulin resistance, immune defect, insulin excels in developing diabetes; regenerative medicine application for metabolic disorders; atypical cases of metabolic diseases modeling in vitro; human mini-organs, organ-on-chip, multi-organ systems.	EU_01, EU_02, EU_06, EU_07
epidemiology and diagnostics of cancer worldwide; diagnostic methods for sporadic and hereditary mutations causing human rare diseases. Genetic epidemiology of rare diseases. Pedigrees.	EU_01, EU_03, EU_06, EU_07
different cancer treatments: surgery, radiotherapy, chemotherapy, modern therapies (specific drugs blocking products of cancer genes, immunotherapies, oncolytic viruses); personalized therapeutic nutrition for metabolic disorders and treatment of rare diseases.	EU_01, EU_04, EU_06

5. Reading list

Wydawnictwa książkowe: fragments provided by the teacher

1. Fred Bunz: Principles of Cancer Genetics, Springer, London, 2016
2. Robert A. Weinberg: The Biology of Cancer, WW Norton & Co, New York, 2014
3. Robert L. Nussbaum, MD, FACP, FACMG, Roderick R. McInnes, CM, MD, PhD, FRS(C), FCAHS, FCCMG and Huntington F Willard, PhD: Thompson and Thompson, Elsevier, Amsterdam, 2015
4. Jules Berman: Rare Diseases and Orphan Drugs, Elsevier, Amsterdam, 2014
5. Saudubray, Jean-Marie, Baumgartner, Matthias, Walter, John: Inborn Metabolic Diseases, Springer, London, 2016
6. Hoffmann, Georg F., Zschocke, Johannes, Nyhan, William L.: Inherited Metabolic Diseases, Springer, London, 2017

Artykuły w czasopismach

1. Barroso I, McCarthy MI. (2019): The Genetic Basis of Metabolic Disease, Cell, Mar 21;177(1):146-161
2. Boughton CK, Hovorka R. (2019): Advances in artificial pancreas systems, Sci Transl Med, Mar 20;11(484).
3. Sudesna Chatterjee et al. (2017): Type 2 diabetes, Lancet, Jun 3;389(10085):2239-2251.

4. Trevor Richter et al. (2015): Rare Disease Terminology and Definitions—A Systematic Global Review: Report of the ISPOR Rare Disease Special Interest Group, Value in Health, Vol. 18, Issue 6, p. 906-914
5. Special Issue - series of articles (2020): DNA damage repair, Nature Reviews, February

III. Additional information

1. Teaching and learning methods and activities to enable students to achieve the intended course learning outcomes (please indicate the appropriate methods and activities with a tick or/and suggest different methods)

Teaching and learning methods and activities	
Lecture with a multimedia presentation	X
Interactive lecture	
Problem – based lecture	
Discussions	
Text-based work	
Case study work	
Problem-based learning	
Educational simulation/game	
Task – solving learning (eg. calculation, artistic, practical tasks)	
Experiential work	
Laboratory work	X
Scientific inquiry method	
Workshop method	
Project work	
Demonstration and observation	
Sound and/or video demonstration	
Creative methods (eg. brainstorming, SWOT analysis, decision tree method, snowball technique, concept maps)	
Group work	

2. Assessment methods to test if learning outcomes have been achieved (please indicate with a tick the appropriate methods for each LO or/and suggest different methods)

Assessment methods	Course learning outcome symbol						
	EU_1	EU_2	EU_3	EU_4	EU_5	EU_6	EU_7
Written exam							
Oral exam							
Open book exam							
Written test	X	X	X	X	X	X	
Oral test							
Multiple choice test							
Project							
Essay							
Report							
Individual presentation							
Practical exam (performance observation)							X
Portfolio							

3. Student workload and ECTS credits

Activity types	Mean number of hours spent on each activity type
Contact hours with the teacher as specified in the study programme	30
Preparation for classes	10
Reading for classes	25
Essay / report / presentation / demonstration preparation, etc.	
Project preparation	
Term paper preparation	
Exam preparation	25
Total hours	90
Total ECTS credits for the course	3

4. Assessment criteria according to AMU in Poznan grade system

Very good (bdb; 5,0): Student has very good knowledge of the entire content taught during classes.
Good plus (+db; 4,5): Student has very good knowledge of the entire content taught during classes with some minor errors.
Good (db; 4,0): Student has good knowledge of the entire content taught during classes.
Satisfactory plus (+dst; 3,5): Student has good knowledge of the entire content taught during classes with some minor errors.
Satisfactory (dst; 3,0): Student has sufficient knowledge of the entire content taught during classes
Unsatisfactory (ndst; 2,0): Student has insufficient knowledge of the entire content taught during classes