SUBJECT CARD

Faculty of Medicine and Health Sciences
Field of studies: Medicine
Form of studies: Full-time
Degree: long-cycle Master's program
Specializations: No specialization
Academic year: 2022/2023

GENETICS	
SUBJECT	**Genetics**
NUMBER OF ECTS POINTS	3
LANGUAGE OF INSTRUCTION	English
TEACHER(S)	Assoc. Professor Anna Sadakierska-Chudy, MD, PhD
PERSON RESPONSIBLE	Assoc. Professor Anna Sadakierska-Chudy, MD, PhD
NUMBER OF HOURS	
LECTURES	30 h
CLASSES	15 h
GENERAL OBJECTIVES	
OBJECTIVE 1	Understanding the principles of inheritance and the contribution of genetic factors in the etiopathogenesis of diseases of various systems. Getting acquainted with specialized dysmorphology nomenclature.
OBJECTIVE 2	Acquainting with diagnostic methods detecting gene and chromosomal mutations responsible for hereditary and neoplastic diseases. Getting to know the basic methods of classic and molecular cytogenetics as well as formulas for writing a karyotype according to rules and nomenclature.
OBJECTIVE 3	Understanding the indications for particular genetic tests in pre- and postnatal diagnostics. Presentation of ethical, moral and legal problems related to genetic diagnosis and counseling.
LEARNING OUTCOMES	
MK1	**Knowledge:** Student knows the basic concepts of genetics.
MK2	**Knowledge:** Student describes phenomena of genetic linkage and gene interaction.
MK3	**Knowledge:** Student describes the normal human karyotype and various types of sex determination.
MK4	**Knowledge:** Student describes the structure of chromosomes and the molecular basis of mutagenesis.

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MK5	**Knowledge:** Student knows the principles of inheritance of various number of traits and quantitative traits as well as independent inheritance of traits and inheritance of non-nuclear genes.
MK6	**Knowledge:** Student knows the genetics of human blood groups and the serological conflict in the Rh system.
MK7	**Knowledge:** Student describes the aberration of autosomes and sex chromosomes that cause diseases, including cancers.
MK8	**Knowledge:** Student knows factors affecting the primary and secondary genetic balance of the population.
MK9	**Knowledge:** Student knows the basics of molecular diagnostics of point mutations and chromosomal (numerical and structural) aberrations responsible for hereditary and acquired diseases, including neoplastic diseases.
MS1	**Skills:** Student analyzes genetic crosses and pedigrees of human traits and diseases, as well as assesses the risk of a child with chromosomal aberrations.
MS2	**Skills:** Student identifies indications for prenatal testing.
MS3	**Skills:** Student decides about the need for cytogenetic and molecular tests.
MS4	**Skills:** Student estimates the risk of disease transmission to the offspring based on family predispositions and the impact of environmental factors.

INTRODUCTORY REQUIREMENTS
[1] Knowledge of the molecular structures of organic compounds that build human body. The ability to characterize the basic cellular processes important for the proper functioning of the human body. **[2]** Knowledge of cytobiology and the basics of genetics and basic genetic mechanisms.

COURSE PROGRAM	DETAILED DESCRIPTION OF THE TOPIC BLOCKS
LECTURE 1	Fundamentals of molecular genetics: structure of nucleic acids, synthesis and organization of DNA, structure and function of genes, expression of genetic information.
LECTURE 2	The structure of the human genome. Genetic variation: mutations and polymorphisms. DNA repair mechanisms.
LECTURE 3	Mendel's laws and models of inheritance. Autosomal dominant and recessive inheritance. A variation on Mendel's laws (gene association and interaction).
LECTURE 4	Sex-linked inheritance. Polygenic inheritance and environmental effects. Non-Mendelian inheritance.
LECTURE 5	Numerical and structural chromosomal aberrations. Methods of chromosome analysis - classical and molecular cytogenetics.

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LECTURE 6	Chromosomal disorders - syndromes caused by numerical and structural aberrations.
LECTURE 7	Epigenetics: DNA methylation, post-translational histone modifications, epigenetic regulation of gene expression, genomic imprinting and X chromosome inactivation. The role of epigenetic in cancer.
LECTURE 8	Pharmacogenomics and personalized medicine: genetic polymorphisms of enzymes involved in drug metabolism and individualized diseases prevention and therapy.
LECTURE 9	Genetic metabolic diseases. Errors in the metabolism of amino acids, carbohydrates and lipids. Lysosomal storage diseases. Heavy metal ions transport disorders. Diagnostics of metabolic defects.
LECTURE 10	Immunogenetics. The genetic origin of antibodies and antigens diversity. Hereditary diseases of the immune system.
LECTURE 11	Cancer genetics (oncogenetics). Suppressor genes, mutator genes and oncogenes. Stages of carcinogenesis. Family cancer predisposition syndromes.
LECTURE 12	Neurogenetic diseases: the influence of genetic factors on nervous system dysfunctions. Molecular diagnosis of neurogenetic diseases.
LECTURE 13	Sex determination in Humans. Chromosomal, genetic and gonadal sex. Classification of gender differentiation disorders.
LECTURE 14	The genetics of infertility. Prenatal and preimplantation diagnostics. Invasive and non-invasive tests. Treatment of fetal defects. Legal and ethical issues in prenatal diagnosis.
LECTURE 15	Treatment of genetic diseases - gene therapy, limitations and perspectives.
CLASS 1	Organization of exercises and requirements for passing the course. Principles of safety regarding work in a molecular laboratory. Collection of material for genetic testing and rules of material handling. DNA isolation from epithelial cells.
CLASS 2	Determination of *ACE* gene insertion/deletion (I/D) polymorphism by PCR method. PCR variants, restriction fragment length analysis and interpretation of results based on case studies.
CLASS 3	Determination of *ACE* gene polymorphism (continuation): agarose gel preparation, the separation of PCR products by electrophoresis and interpretation of the results. Genetic tasks with elements of population genetics. Prediction of the frequency of alleles and genotypes.
CLASS 4	Classical cytogenetics - a type of biological material used in cytogenetic research, chromosome staining techniques. Karyotype analysis and description according to the ISCN nomenclature.

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CLASS 5	Genetic counseling - family history interpretation and risk assessment. Practical use of the database.
DIDACTIC METHODS (APPLIED)	
	Lectures, Laboratory classes, Discussion, Solving tasks, Case studies.
STUDENTS WORKLOAD	
NUMBER OF HOURS UNDER SUPERVISION	45 hours
NUMBER OF PREPARATION HOURS	Preparation for classes: 10 hours Processing of results: 5 hours Preparation for the exam: 30 hours
TOTAL NUMBER OF HOURS FOR THE COURSE	90 hours
CONDITIONS FOR COURSE COMPLETION	
[1] Lectures and classes are obligatory.	
[2] Condition to pass the lab classes is: **100% presence** and active participation in them, and passing the colloquium.	
[3] It is possible to do classes with another students group only in exceptional, justified case, only if agreed with the teacher in advance.	
[4] A colloquium in written form (test) will take place two weeks after the end of the entire exercise cycle.	
[5] In the case of failing the colloquium, the retake written colloquium will take place not later than 1 week after the date of the first colloquium.	
[6] The colloquium will cover the material from the lab classes. To pass the colloquium student should get min. 60% of the entire pool of points possible to obtain.	
[7] Passing the classes (in accordance with point 2) is necessary for admission to the final exam.	
[8] The final written exam will cover material presented in both lectures and labs/exercises.	
[9] The exam will consist of open-ended questions and genetic tasks. To pass the final exam student should gain min. 55% of the entire pool of points.	
[10] The form and conditions for passing the retake exam will be the same as above (point 9).	
[11] The exam grade will be the final grade for the course.	

GENETICS	
METHODS OF ASSESMENT	
IN TERMS OF KNOWLEDGE	Test, open-ended questions, task-solving.
IN TERMS OF SKILLS **FORMATIVE**	Drawing a pedigree based on a case study. Estimating the risk of genetic disease in offspring and making the decision to perform genetic tests (prenatal and postnatal) based on family predispositions. Description of the karyotype based on the cytogenetic test result.
SUMMATIVE **(I & II TERMS)**	**I term (EXAM):** written form, open-ended questions and task-solving. **II term (RETAKE EXAM):** written form, open-ended questions and task-solving.
GRADING SCALE	
3,0 (SATISFACTORY)	the range **55 - 65 points**
3,5 (SATISFACTORY PLUS)	the range **66 - 71 points**
4,0 (GOOD)	the range **72 - 82 points**
4,5 (GOOD PLUS)	the range **83 - 90 points**
5,0 (VERY GOOD)	the range **91 - 100 points**
BASIC LITERATURE	
[1] Jorde LB, Carey JC, Bamshad MJ. — *Medical Genetics*, Canada, 2015, Elsevier [5th edition].	
SUPPLEMENTARY LITERATURE	
[1] G.I. Hickey, H.L. Fletcher, and P. Winte — *BIOS Instant Notes in Genetics*, USA, 2007, Taylor & Francis Group [3rd edition]; **[2]** Tobias ES, Connor JM, Ferguson-Smith M. – *Essential Medical Genetics*, Singapore, 2011, Wiley-Blackwell [6th edition].	