The schedule of lectures and practical lessons for Medical genetics

Study program **Medicine,** academic year 2023-2024

Date	Lectures	Date	Practical lessons
05.02. - 09.02.	Introduction. The genetic apparatus of the human cell. Base of heredity and variability. Heredity and the environment in the manifestation of normal and pathological traits in humans.	05.02. - 09.02.	Genetic apparatus - components, role in the storage, transmission, and realization of GI. Heredity and variability. Mutations - types, origin, phenotypic consequences.
12.02. - 16.02.	Chromosomes - the morphological base of heredity and variability. Characteristic of the human karyotype. Peculiarities of X and Y chromosomes. Chromosomes polymorphisms. Uniparental disomy. Classification of chromosomal abnormalities.	12.02. - 16.02.	Assessment of normal karyotype and structural, numerical variations. Nomenclature of normal and pathological variations of the karyotype.
19.02. - 23.02.	Numerical and structural chromosomal abnormalities. Causes, mechanisms of production and their consequences: reproductive abnormalities, birth defects and mental retardation	19.02. - 23.02.	Cytogenetic methods - diversity, indications, and limits.
26.02. - 01.03.	Chromosomal syndromes - diversity, causes, manifestations and cytogenetic diagnosis.	26.02. - 01.03.	Numerical and structural chromosomal abnormalities - types, production mechanisms, consequences.
04.03. - 07.03.	Human genes - organization, properties, functions, diversity, examples. Gene-genotype-phenotype relationship.	04.03. - 07.03.	Autosomal and gonosomal chromosomal syndromes - cytogenetic characteristics, major phenotypic manifestations, diagnosis, prognosis and prevention.
11.03. - 15.03.	Gene mutations - mechanisms, nomenclature, consequences at molecular, cellular and organism level. Pathological vs positive vs neutral gene mutations.	11.03. - 15.03.	**Concluding test 1.** Chromosomes and chromosomal diseases.
18.03. - 22.03.	Phenomena in the expression of human genes. Parental fingerprinting of human genes and monoallelic expression. Epigenetic changes and their role in human pathology.	18.03. - 22.03.	Human genes - diversity, properties, functions. Autosomal genes and X-linked genes. Allelic and non-allelic genes. Linkage groups. Haplotypes.
25.03. - 29.03.	Methods of studying genes. Direct and indirect gene testing. Advantages and limitations of human gene testing.	25.03. - 29.03.	Gene mutations - classification, nomenclature, consequences. AD, AR, XD, XR, mitochondrial monogenic diseases.
01.04. - 05.04.	Peculiarities of normal vs. pathological hereditary traits. Methods for assessing the genetic nature of human diseases. Genealogical, twins, linkage, genomic studies.	01.04. - 05.04.	Principles of PCR, RFLPs and dideoxy sequencing techniques. Prenatal and postnatal testing of human genes.
08.04. - 12.04.	Mendelian (AD, AR, AD, XR) and non-Mendelian (mitochondrial, fingerprinting) monogenic diseases, common polygenic diseases.	08.04. - 12.04.	Mendelian monogenic traits. Heredity of ABO, Rh, MN and Xg blood groups. Allelic and non-allelic interactions. Genetics of HDNB.
15.04. - 19.04.	Developmental genetics. Genetic basis of congenital anomalies.	15.04. - 19.04.	Non-mendelian monogenic traits. Allelic and non-allelic heterogeneity. Complete and incomplete penetration. Variable expressivity. Anticipation.
22.04. - 26.04.	The genetics of sex. Major masculinizing and feminizing genes. Sex development disorders.	22.04. - 26.04.	**Concluding test 2.** Human genes.
29.04. - 03.05.	Immunogenetics. Genetic mechanisms of Ac and TcR diversity. Genetic immunodeficiencies.	29.04. - 03.05.	Study of normal and pathological hereditary traits. Methods for confirming the genetic nature of human diseases.
14.05. - 17.05.	Cancer - a genetic disease of somatic cells. Mutations of proto-oncogenes and GST in carcinogenesis.	14.05. - 17.05.	Genealogical analysis of monogenic diseases. Assessment of the type of transmission and calculation of the risk of recurrence
20.05. - 24.05.	Genetic counseling in the prevention of genetic diseases. Principles of gene therapies in the etiological treatment of genetic diseases. Pharmacogenomic perspectives.	20.05. - 24.05.	**Concluding test 3**. Peculiarities of hereditary traits. Diversity, etiology, and prophylaxis of genetic diseases. Prenatal and postnatal genetic diagnosis - indications and limits.
	Total: 30 hours	**Total: 45 hours**	

30.01.2024

Head of Department Dr. Igor Cemortan