

Summary of the working program of the academic discipline

«FUNDAMENTALS OF MEDICAL GENETICS»

General Educational Program of higher education (specialist's degree programs)

31.05.01 GENERAL MEDICINE

Department: **BIOLOGY**

1. The purpose of mastering the discipline (*participation in the formation of relevant competencies – specify the codes*):

UC-1. Able to carry out a critical analysis of problem situations based on a systematic approach, develop an action strategy

2. Position of the academic discipline in the structure of the General Educational Program (GEP).

2.1. The discipline **MEDICAL GENETICS** refers to the core part (*or the part formed by the participants of educational relations*) of Block 1 of GEP HE (Academic discipline index).

The discipline is taught in 3 semester/2 year of study.

3. Deliverables of mastering the academic discipline and metrics of competence acquisition

Mastering the discipline aims at acquiring the following universal (UC) competencies

№	Competence code	The content of the competence (or its part)	Code and name of the competence acquisition metric	As a result of mastering the discipline, the students should:		
				know	be able to	possess
1.	UC-1.	Able to carry out a critical analysis of problem situations based on a systematic approach, develop an action strategy	1.1 methods of critical analysis and evaluation of modern scientific achievements; basic principles of critical analysis 1.2 gain new knowledge based on analysis, synthesis, etc.; collect data on complex scientific problems related to the professional field; search	- principles of analysis of the elements of the information received (identified symptoms, syndromes, pathological changes) as a result of examination of a patient with a hereditary disease; • principles of synthesis of the received information (identified symptoms, syndromes, pathological changes) for making	- analyze the information received, make an independent decision - compile pedigrees using standard notation, analyze pedigrees; - explain the causes and possible mechanisms of the birth of children with chromosomal diseases; - methods of studying	- skills in analyzing scientific literature and official statistical reviews, preparing abstracts, reviews on current and modern scientific issues in the field of hereditary pathology. - skills of screening-evaluation of the results of laboratory and instrumental research

			<p>for information and solutions based on action, experiment and experience</p>	<p>preliminary diagnosis</p> <ul style="list-style-type: none"> - the main types of inheritance, clinical symptoms and syndromes, the nature of the course and outcomes of the most common hereditary diseases - prognosis for life, working capacity and social adaptation in major hereditary diseases. - the main directions of prevention of hereditary diseases. - the main methods of laboratory and instrumental diagnostics necessary for the verification and formulation of the diagnosis of the most common hereditary diseases (genetic, biochemical, morphological foundations of the development of hereditary pathology); - indications for referral to various instrumental and laboratory methods • - the main risk factors for the development of diseases and their correction 	<p>human heredity (cytogenetic method, biochemical method, genealogical method, twin method; the principle of DNA sequencing)</p> <ul style="list-style-type: none"> - interpret the most significant changes in the results of laboratory and instrumental research methods for the diagnosis of hereditary diseases, namely: general and biochemical blood tests, cytogenetic methods, methods of direct DNA diagnostics. - apply modern information technologies to obtain information concerning the diagnosis and treatment of hereditary diseases - identify hereditary diseases - to organize 	<p>methods and identification of those changes that require referral of the patient to a geneticist.</p>
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				<ul style="list-style-type: none"> - the main components of a healthy lifestyle. - principles of the organization of programs for the diagnosis of hereditary diseases - forms and methods of organization of hygienic education and upbringing of the population. - the methodology of medical and genetic counseling 	<ul style="list-style-type: none"> work on the formation of motivation among the population, patients and their family members aimed at preserving and strengthening their health and the health of others. - evaluate the importance of lifestyle for the preservation of human health and plan your life activities based on knowledge about a healthy lifestyle 	
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4. Volume of the academic discipline and types of academic work

Total labor intensity of the discipline is 1 CU (36 AH)

Type of educational work	Labor intensity		Labor intensity (AH) in semesters
	volume in credit units (CU)	volume in academic hours (AH)	
Classroom work, including	0,61	22	
Lectures (L)	0,17	6	6
Laboratory practicum (LP)*	-	-	-
Practicals (P)	0,44	16	16
Seminars (S)	-	-	-
Student's individual work (SIW)	0,39	14	14
Mid-term assessment	-	-	-
credit/exam (<i>specify the type</i>)	-	-	-
TOTAL LABOR INTENSITY	1	36	36

5. Sections of the academic discipline and competencies that are formed

№	Competence code	Section name of the discipline	The content of the section in teaching units
1.	UC-1.	<p>1. Molecular genetics is the basis of medical genetics. Genetic passport. Epigenetics. Ethnogenomics.</p>	<p>Genomics. Proteomics. The human genome as the scientific basis of predictive medicine. Genetic passport. Genomic imprinting is an epigenetic system of gene regulation. Mitochondrial diseases. Genomics and genomic technologies. New projects for the study of the human genome. Genetic polymorphism. Ethnogenomics. Genomics and the creation of new drugs. The concept of drug metabolic safety. Pharmacogenetics.</p>
		<p>2. Methods of studying human genetics. Monogenic, chromosomal and genomic diseases.</p>	<p>Genealogical method of human genetics research. Compilation of pedigrees. Twin method: determination of kinship coefficient; kinship coefficients for different pairs of relatives; determination of concordance Population-statistical method of research in medical genetics. The importance of studying the frequencies of genes and genotypes in a population to obtain information about the frequency of heterozygosity. Biochemical method of human genetics research. Cytogenetic research methods in medical genetics. Standard karyotyping. Fluorescent in situ hybridization (FISH method). Multicolored FISH methods. Examples of application in clinical practice. Monogenic diseases. Types of inheritance. Classification of monogenic diseases. Metabolic diseases: amino acid, carbohydrate, lipid, purine, porphyrin metabolism. Chromosomal aberrations. Classification. The most common pathology: Down syndrome, Shereshevsky-Turner, Klinefelter.</p>
		<p>3. Congenital diseases and malformations.</p>	<p>Congenital diseases and malformations. Defects in monogenic and chromosomal diseases. Defects caused by endocrine, hormonal and metabolic disorders of the mother. Vices of exogenous origin. Multifactorial defects. The role of physical, chemical and biological factors in the origin of congenital developmental disorders.</p>
		<p>4. Methods of diagnosis of hereditary human pathologies and medical and genetic counseling.</p>	<p>Methods of DNA diagnostics. Use in clinical practice.</p> <ul style="list-style-type: none"> • analysis of restriction fragment length polymorphism • analysis of polymorphism of mini- and microsatellite sequences • polymerase chain reaction • analysis of conformational polymorphism of single-stranded DNA • DNA sequencing methods (dideoxy-Sanger method, fluorochromic staining, chemical cleavage) • hybridization of nucleic acids with allele-specific probes. <p>Applied aspects of the application of methods of molecular genetics and DNA diagnostics in clinical medicine. The method of DNA comets in the assessment of the genotoxic effect of environmental factors. Tasks of medical and genetic counseling and indications for referral of patients and their families to medical and genetic counseling. Methods of medical and genetic counseling. The main clinical-genetic and research methods.</p>