

*Summary of the working program of the academic discipline*

**« BASICS OF CLINICAL GENETICS »**

General Educational Program of higher education 31.05.01 general medicine

Department: Hospital pediatrics

**1. The purpose of mastering the discipline:** participation in the formation of the following universal (UC), general professional (GPC) and professional competencies (PC): UC-1, GPC-5, GPC-7, GPC-10, PC-5, PC-6, PC-7

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

**2.1.** The discipline refers to the core part of Block 1 of GEP HE 31.05.01 general medicine.

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

Mastering the discipline aims at acquiring the following universal (UC) or/and general professional (GPC) or/and professional (PC) 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	IUC-1.1. Knows: methods of critical analysis and evaluation of modern scientific achievements; basic principles of critical analysis IUC-1.2. Be able to: gain new knowledge based on analysis, synthesis, etc.; collect data on complex scientific problems related to the professional field; search for information and solutions based on action, experiment and experience IUC-1.3. Has practical experience: researching the problem of professional activity using analysis, synthesis and other methods of intellectual activity;	methods of critical analysis and evaluation of modern scientific achievements; basic principles of critical analysis	gain new knowledge based on analysis, synthesis, etc.; collect data on complex scientific problems related to the professional field; search for information and solutions based on action, experiment and experience	Has practical experience: researching the problem of professional activity using analysis, synthesis and other methods of intellectual activity; developing an action strategy to solve professional problems

			developing an action strategy to solve professional problems			
2.	GPC-5	Able to assess morphofunctional, physiological conditions and pathological processes in the human body to solve professional problems	<p>IGPC-5.1. Knows: anatomy, histology, embryology, topographic anatomy, physiology, pathological anatomy and physiology of human organs and systems.</p> <p>IGPC-5.2. Be able to: evaluate the basic morphological and functional data, physiological conditions and pathological processes in the human body</p> <p>IGPC-5.3. Has practical experience in: assessment of basic morphological and functional data, physiological conditions and pathological processes in the human body when solving professional problems</p>	anatomy, histology, embryology, topographic anatomy, physiology, pathological anatomy and physiology of human organs and systems	evaluate the basic morphological and functional data, physiological conditions and pathological processes in the human body	Has practical experience in: assessment of basic morphological and functional data, physiological conditions and pathological processes in the human body when solving professional problems
3.	GPC-7	Able to prescribe treatment and monitor its effectiveness and safety	<p>IGPC-7.1. Knows: methods of drug and non-drug treatment, medical indications for the use of medical devices for the most common diseases; groups of drugs used to provide medical care in the treatment of the most common diseases; the mechanism of their action, medical indications and contraindications to the prescription; compatibility, possible complications, side effects, adverse reactions, including serious and</p>	methods of drug and non-drug treatment, medical indications for the use of medical devices for the most common diseases; groups of drugs used to provide medical care in the treatment of the most common diseases; the mechanism of their action, medical indications and contraindications to the prescription; compatibility,	develop a treatment plan for patients with the most common diseases in accordance with the procedures for providing medical care, clinical guidelines taking into account the standards of medical care; prevent or eliminate complications, side effects, adverse reactions,	Has practical experience in: developing a treatment plan for patients with the most common diseases in accordance with the current procedures for the provision of medical care, clinical guidelines, taking into account the standards of medical care; provision of emergency and urgent medical care to patients with the most

			<p>unforeseen; features of medical care in emergency forms. IGPC-7.2. Be able to: develop a treatment plan for patients with the most common diseases in accordance with the procedures for providing medical care, clinical guidelines taking into account the standards of medical care; prevent or eliminate complications, side effects, adverse reactions, including unforeseen ones, resulting from diagnostic or therapeutic manipulations, the use of drugs and (or) medical devices, non-drug treatment IGPC-7.3. Has practical experience in: developing a treatment plan for patients with the most common diseases in accordance with the current procedures for the provision of medical care, clinical guidelines, taking into account the standards of medical care; provision of emergency and urgent medical care to patients with the most common diseases in accordance with the current procedures for providing medical care, clinical guidelines, taking into account the standards of medical care; selection and prescription of</p>	<p>possible complications, side effects, adverse reactions, including serious and unforeseen; features of medical care in emergency forms</p>	<p>including unforeseen ones, resulting from diagnostic or therapeutic manipulations, the use of drugs and (or) medical devices, non-drug treatment</p>	<p>common diseases in accordance with the current procedures for providing medical care, clinical guidelines, taking into account the standards of medical care; selection and prescription of medicines, medical devices with the most common diseases for the treatment of the most common diseases of children and adults in accordance with the current procedures for providing medical care, clinical recommendations, taking into account the standards of medical care; evaluation of the effectiveness and safety of the use of drugs, medical devices and non-drug treatment of patients with the most common diseases; selection and prescription of non-drug treatment for patients with the most common diseases in accordance with the procedures for providing medical care, clinical guidelines, taking into</p>
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			the patient's life in an emergency form; the use of medicines and medical devices in the provision of medical care in an emergency form.			
4	GPC-10	Able to understand the principles of modern information technologies and use them to solve the tasks of professional activity	IGPC-10.1. Knows: the capabilities of reference information systems and professional databases; methods of information retrieval, information and communication technologies; modern medical and biological terminology; fundamentals of information security in professional activities. IGPC-10.2. Be able to: Apply modern information and communication technologies to solve the tasks of professional activity; carry out an effective search for information necessary to solve the tasks of professional activity using reference systems and professional databases; use modern medical and biological terminology; master and apply modern information and communication technologies in professional activity, taking into account the basic requirements of information security. IGPC-10.3. Has practical experience in the use of modern information and	the capabilities of reference information systems and professional databases; methods of information retrieval, information and communication technologies; modern medical and biological terminology; fundamentals of information security in professional activities	Apply modern information and communication technologies to solve the tasks of professional activity; carry out an effective search for information necessary to solve the tasks of professional activity using reference systems and professional databases; use modern medical and biological terminology; master and apply modern information and communication technologies in professional activity, taking into account the basic requirements of information security	Has practical experience in the use of modern information and bibliographic resources, the use of special software and automated information systems to solve standard tasks of professional activity, taking into account the basic requirements of information security

			<p>bibliographic resources, the use of special software and automated information systems to solve standard tasks of professional activity, taking into account the basic requirements of information security</p>			
5	PC-5	<p>Able to collect complaints, anamnesis of the patient's life and illness, conduct a complete physical examination of the patient (examination, palpation, percussion, auscultation), formulate a preliminary diagnosis and draw up a plan for laboratory and instrumental examinations of the patient</p>	<p>IPC-5.1. Knows: the legislation of the Russian Federation in the field of health care, regulations and other documents that determine the activities of medical organizations and healthcare workers; method of collecting complaints, anamnesis of life and disease of the patient; a technique for a complete physical examination of the patient (examination, palpation, percussion, auscultation); etiology, pathogenesis and pathomorphology, clinical aspect, differential diagnosis, clinical features, complications and outcomes of diseases of internal organs; patterns of functioning of a healthy human body and mechanisms for ensuring health from the standpoint of the theory of functional systems; features of the regulation of the functional systems of the human body in pathological processes; methods of laboratory and</p>	<p>the legislation of the Russian Federation in the field of health care, regulations and other documents that determine the activities of medical organizations and healthcare workers; method of collecting complaints, anamnesis of life and disease of the patient; a technique for a complete physical examination of the patient (examination, palpation, percussion, auscultation); etiology, pathogenesis and pathomorphology, clinical aspect, differential diagnosis, clinical features, complications and outcomes of diseases of internal organs; patterns of functioning of a healthy human body and mechanisms for ensuring health from the standpoint of the theory of</p>	<p>collect complaints, anamnesis of life and disease of the patient and analyze the information received; conduct a complete physical examination of the patient (examination, palpation, percussion, auscultation) and interpret its results; determine the sequence of volume, content and sequence of diagnostic measures incl. modern digital technologies</p>	

			instrumental examinations for assessing the state of health, medical indications for conducting investigations, rules for interpreting their results. IPC-5.2. Be able to: collect complaints, anamnesis of life and disease of the patient and analyze the information received; conduct a complete physical examination of the patient (examination, palpation, percussion, auscultation) and interpret its results; determine the sequence of volume, content and sequence of diagnostic measures incl. modern digital technologies	functional systems; features of the regulation of the functional systems of the human body in pathological processes; methods of laboratory and instrumental examinations for assessing the state of health, medical indications for conducting investigations, rules for interpreting their results		
6	PC-6	Able to send a patient to laboratory, instrumental examination, to a consultation with specialist doctors if there are medical indications in accordance with the current procedures for the provision of medical care, clinical recommendations (treatment protocols) on providing medical care taking into account the standards of medical care,	IPC-6.1. Knows: general issues of organizing medical care for the population, methods of laboratory and instrumental examinations to assess the state of health, medical indications for conducting examinations, rules for interpreting their results; procedures for the provision of medical care, clinical recommendations (treatment protocols) on the provision of medical care, standards of medical care. IPC-6.2. Be able to: justify the need and scope of laboratory examination of the	general issues of organizing medical care for the population, methods of laboratory and instrumental examinations to assess the state of health, medical indications for conducting examinations, rules for interpreting their results; procedures for the provision of medical care, clinical recommendations (treatment protocols) on the provision of medical care, standards of medical care	justify the need and scope of laboratory examination of the patient; justify the need and scope of instrumental examination of the patient; justify the need to refer the patient for consultations with specialist doctors; determine medical indications for the provision of emergency, including emergency specialized, medical care	

		and also refer the patient for providing specialized medical care in an inpatient setting or in a day hospital if there are medical indications in accordance with the current procedures for the medical care delivery, clinical recommendations (treatment protocols) on the medical care delivery taking into account the standards of medical care	patient; justify the need and scope of instrumental examination of the patient; justify the need to refer the patient for consultations with specialist doctors; determine medical indications for the provision of emergency, including emergency specialized, medical care			
7	PC-7	Able to: make a differential diagnosis with other diseases/conditions including emergencies, establish a diagnosis taking into account the current international statistical classification of diseases and related health problems (ICD)	IPC-7.1. Knows: etiology, pathogenesis and pathomorphology, clinical aspect, differential diagnosis, clinical features, complications and outcomes of diseases of internal organs; methods of laboratory and instrumental examinations for assessing the state of health, medical indications for conducting examinations, rules for interpreting their results; ICD. IPC-6.2. Be able to: analyze the results of the patient's examination, if necessary, justify and plan the scope of additional examinations; interpret the results of collecting	etiology, pathogenesis and pathomorphology, clinical aspect, differential diagnosis, clinical features, complications and outcomes of diseases of internal organs; methods of laboratory and instrumental examinations for assessing the state of health, medical indications for conducting examinations, rules for interpreting their results; ICD	analyze the results of the patient's examination, if necessary, justify and plan the scope of additional examinations; interpret the results of collecting information about the patient's disease; interpret the data obtained during the laboratory examination of the patient; interpret the data obtained during the instrumental examination of the patient; interpret the data obtained during consultations	



		information about the patient's disease; interpret the data obtained during the laboratory examination of the patient; interpret the data obtained during the instrumental examination of the patient; interpret the data obtained during consultations of the patient by medical specialists; to carry out differential diagnostics of diseases of internal organs from other diseases		of the patient by medical specialists; to carry out differential diagnostics of diseases of internal organs from other diseases	
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#### 4. Volume of the academic discipline and types of academic work

Total labor intensity of the discipline is 1,0 CU (36 AH)

Type of educational work	Labor intensity		Labor intensity (AH) in semesters	
	volume in credit units (CU)	volume in academic hours (AH)	5 <sup>th</sup> semester	6 <sup>th</sup> semester
Classroom work, including				
Lectures (L)	0,1	6		6
Laboratory practicum (LP)*				
Practicals (P)				
Clinical Practice (CP)	0,5	16		16
Seminars (S)				
Student's individual work (SIW)	0,4	14		14
Mid-term assessment				
credit				credit
<b>TOTAL LABOR INTENSITY</b>	<b>1,0</b>	<b>36</b>		<b>36</b>

#### 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 GPC-5 GPC-7 GPC-10 PC-5 PC-6	Introduction to clinical genetics. Personalized diagnosis and treatment of hereditary	Medical genetics as a science. Clinical genetics, tasks. General patterns of pathogenesis of hereditary diseases. Classification and modern diagnosis of hereditary diseases. Hereditary pathology and methods of its study. genealogical method. The concept of personalized medicine. Personalized diagnosis and treatment of hereditary diseases. Determining the tactics of

	PC-7	diseases.	managing a patient with hereditary diseases.
2.	UC-1 GPC-5 GPC-7 GPC-10 PC-5 PC-6 PC-7	Modern genetic research methods. Medical genetic counseling.	Modern genetic research methods, indications, interpretation of results. Neonatal screening. Tasks and indications for medical genetic counseling. Principles and stages of counseling.
3.	UC-1 GPC-5 GPC-7 GPC-10 PC-5 PC-6 PC-7	Chromosomal diseases. Prenatal diagnosis of congenital and hereditary diseases.	Definition and modern classification. Factors that cause human chromosomal diseases. Characteristics of the main human chromosomal diseases. Prevention of chromosomal diseases. Modern methods of diagnosing chromosomal diseases. Modern methods of treatment and rehabilitation of patients with chromosomal diseases. Prenatal diagnosis of congenital and hereditary diseases. Prevention of congenital pathology in women at high risk.