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

out a critical analysis of problemmethods of critical analysiscritical and evaluation of modernknowledge based on analysis,situations based on a systematic approach, develop action strategyscientific principles of critical analysisachievements; basic principles of critical analysisscientific achievements; basic principles of critical analysisanalysis, principles of critical analysisprinciples critical analysisdevelop action strategyIUC-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 experiencemethods of critical analysisanalysis scientific problems related to the professional field; search for information and solutions based on action, experiment and experienceanalysisknowledge based on action, experiment and experience		olessional (GPC) of/and			1 1 1	• .1 . 1 .
Ne etence code the competence (or its part) the competence acquisition metric know be able to 1. UC-1 Able to carry out a critical analysis of problem IUC-1.1. Knows: methods of critical analysis methods of critical analysis gain new knowledge H knowledge based on a systematic approach, develop an action strategy systematic 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-1.2. Be able to: methods of critical analysis collect data on complex scientific problems related to the professional field; search for information and solutions based on action, experiment and experience information action, experiment and experience methods critical analysis information action, experiment and experience				As a result of ma	v	ine, the students
code(or its part)acquisition metricknowbe able to1.UC-1Able to carry out a critical analysis of problem situationsIUC-1.1.Knows: methods of critical analysismethods of critical analysis and evaluation of modern scientific achievements; basic principles of critical analysisgain new knowledge based on a systematic approach, develop an action strategyIUC-1.2.Be able to: methods of critical analysisgain new knowledge basic principles of critical analysisH knowledge basic principles of critical analysis1.UC-1UC-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 experiencemethods of critical analysisgain new knowledge problems related to the information and solutions based on action, experiment and experiencemethods collect data on complex scientific problems related to the professional field; search for information and solutions based on action, experiment and experiencemethods collect data on complex scientific problems related to the professional field; search for information and solutions based on action, experiment and experienceMethods collect action, experiment and experienceUC-1.3.Has practical experience:H		L			should:	
1.UC-1Able to carry out a critical analysis of problem situationsIUC-1.1.Knows: methods of critical analysismethods of critical analysis and evaluation of modern scientific achievements; basic principles of critical analysisgain new knowledge basic principles of critical analysisH knowledge basic principles of critical analysis1.UC-1Able to carry out a critical analysis of problem systematic approach, develop an action strategyIUC-1.1.Knows: methods of critical analysismethods of critical analysis analysisgain new hknowledge 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 experiencemethods of critical analysisgain new hknowledge 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 experiencemethods of critical analysisgain new hends collect data on complex scientific problems related to the professional field; search for information and solutions based on action, experiment and experienceHIII	Nº	1	-	1	1	
out a critical analysis of problemmethods of critical analysiscritical and evaluation of modernknowledge based on achievements;situationsscientific principles of critical approach, action strategyachievements; principles of critical analysisachievements; basic principles of critical analysissolutions problem critical analysiscritical analysis and evaluation of modern scientific achievements; basic principles of critical analysiscollect data on accion strategydevelop action strategyIUC-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:methods of critical analysisanalysis critical analysis solutions based on action, experiment and experiencemodern scientific problems related to the professional field; search for action, experiment and experiencemethods of critical and solutions action, experiment and experiencemodern scientific problems action, experiment and experience		(or its part)	acquisition metric	KNOW	be able to	possess
researching the problem of professional activity using analysis, synthesis and other methods of intellectual activity;	1.	out a critical analysis of problem situations based on a systematic approach, develop an	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	critical analysis and evaluation of modern scientific achievements; basic principles of	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	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 morphofunction al, physiological conditions and pathological processes in the human body to solve professional problems	problemsIGPC-5.1.Knows:anatomy, histology,embryology,topographicanatomy, physiology,pathological anatomyand physiology ofhuman organs andsystems.IGPC-5.2.Be ableto: evaluate the basicmorphological andfunctional data,physiologicalconditions andpathologicalprocesses in thehuman bodyIGPC-5.3.Haspractical experiencein: assessment ofbasic morphologicalconditions andpathologicalconditions andpathologicalprocesses in thehuman bodyIGPC-5.3.Haspractical experiencein: assessment ofbasic morphologicalconditions andpathologicalprocesses in thehuman body whensolving professionalproblems	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	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	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

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	unforeseen; features	possible	including	common diseases
	of medical care in	complications,	unforeseen	in accordance
	emergency forms.	side effects,	ones, resulting	with the current
	IGPC-7.2. Be able	adverse reactions,	from	procedures for
	to: develop a	including serious	diagnostic or	providing
	treatment plan for	and unforeseen;	therapeutic	medical care,
	patients with the	features of	manipulations,	clinical
	most common	medical care in	the use of	guidelines,
	diseases in	emergency forms	drugs and (or)	taking into
	accordance with the	emergency forms	medical	account the
	procedures for		devices, non-	standards of
	*		,	
	1 0		drug treatment	,
	care, clinical			selection and
	guidelines taking into			prescription of
	account the standards			medicines,
	of medical care;			medical devices
	prevent or eliminate			with the most
	complications, side			common diseases
	effects, adverse			for the treatment
	reactions, including			of the most
	unforeseen ones,			common diseases
	resulting from			of children and
	diagnostic or			adults in
	therapeutic			accordance with
	manipulations, the			the current
	use of drugs and (or)			procedures for
	medical devices,			providing
	non-drug treatment			medical care,
	IGPC-7.3. Has			clinical
	practical experience			recommendation
	in: developing a			s, taking into
	treatment plan for			account the
	patients with the			standards of
	most common			medical care;
	diseases in			evaluation of the
				effectiveness and
	accordance with the			
	current procedures			safety of the use
	for the provision of			of drugs, medical
	medical care, clinical			devices and non-
	guidelines, taking			drug treatment of
	into account the			patients with the
	standards of medical			most common
	care; provision of			diseases;
	emergency and			selection and
	urgent medical care			prescription of
	to patients with the			non-drug
	most common			treatment for
	diseases in			patients with the
	accordance with the			most common
	current procedures			diseases in
	for providing			accordance with
	medical care, clinical			the procedures
	guidelines, taking			for providing
	into account the			medical care,
	standards of medical			clinical
	care; selection and			guidelines,
	prescription of			taking into
				inition initio

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	medicines, medical		account the
	devices with the		standards of
	most common		medical care;
	diseases for the		prevention and
	treatment of the most		treatment of
	common diseases of		complications,
	children and adults in		side effects,
	accordance with the		adverse
	current procedures		reactions,
	for providing		including
	medical care, clinical		unforeseen ones,
	recommendations,		resulting from
	taking into account		diagnostic or
	the standards of		therapeutic
	medical care;		manipulations,
	evaluation of the		the use of drugs
	effectiveness and		and (or) medical
	safety of the use of		devices, non-
	-		,
	6		drug treatment;
	devices and non-drug		providing
	treatment of patients		medical care to
	with the most		patients with
	common diseases;		sudden acute
	selection and		diseases,
	prescription of non-		conditions,
	drug treatment for		exacerbation of
	patients with the		chronic diseases
	most common		without obvious
	diseases in		signs of a threat
	accordance with the		to the patient's
			11.0
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	providing medical		emergency form;
	care, clinical		the use of
	guidelines, taking		medicines and
	into account the		medical devices
	standards of medical		in the provision
	care; prevention and		of medical care
	treatment of		in an emergency
	complications, side		form
	effects, adverse		
	reactions, including		
	unforeseen ones,		
	resulting from		
	diagnostic or		
	therapeutic		
	manipulations, the		
	use of drugs and (or)		
	medical devices,		
	non-drug treatment;		
	providing medical		
	care to patients with		
	sudden acute		
	diseases, conditions,		
	exacerbation of		
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	chronic diseases		
	chronic diseases without obvious signs of a threat to		

		the patient's life in an emergency form; the use of medicines and medical devices in the provision of medical care in an			
GPC- 10	Able to understand the principles of modern information technologies and use them to solve the tasks of professional activity	emergency form.IGPC-10.1.Knows:the capabilities ofreference informationsystemsandprofessionaldatabases;databases;methodsofinformationretrieval, informationandcommunicationtechnologies;modernmedicalandbiologicalterminology;fundamentalsofinformationsecurityinprofessionalactivities.IGPC-10.2.IGPC-10.2.Be ableto:Applymodernandcommunicationandcommunicationandcommunicationandcommunicationandcommunicationforinformationandcommunicationforinformationandcommunicationforinformationandcommunicationforinformationandnecessaryto solvethetasksofprofessionalactivityusingusingreferencesystemsandprofessionalactivityusingreferencesystemsandprofessionalactivityusingreferencesystemsandprofessionalactivityusingreferencesystemsandprofessionalactivityusingreference<	the capabilities of reference information systems and professional databases; methods of information and communication technologies; modern medical and biological terminology; fundamentals of information security in professional activities	Apply modern information and communicatio n 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 communicatio n 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

			hibliographia			
			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			
-		A11 · 11	information security	4 1 1 1 2 2	11	
5	PC-5	Able to collect	IPC-5.1. Knows: the	the legislation of	collect	
•		complaints,	legislation of the	the Russian	complaints,	
		anamnesis of	Russian Federation	Federation in the	anamnesis of	
		the patient's	in the field of health	field of health	life and	
		life and illness,	care, regulations and	care, regulations	disease of the	
		conduct a	other documents that	and other	patient and	
		complete	determine the	documents that	analyze the	
		physical	activities of medical	determine the	information	
		examination of	organizations and	activities of	received;	
		the patient	healthcare workers;	medical	conduct a	
		(examination,	method of collecting	organizations and	complete	
		palpation,	complaints,	healthcare	physical	
		percussion,	anamnesis of life and	workers; method	examination of	
		auscultation),	disease of the	of collecting	the patient	
		formulate a	patient; a technique	complaints,	(examination,	
		preliminary	for a complete	anamnesis of life	palpation,	
		diagnosis and	physical examination	and disease of the	percussion,	
		draw up a plan	of the patient	patient; a	auscultation)	
		for laboratory and instrumental	(examination,	technique for a	and interpret its results;	
		examinations of	palpation,	complete physical examination of	its results; determine the	
			percussion,			
		the patient	auscultation); etiology,	the patient (examination,	sequence of volume,	
			•••	· ·	content and	
			pathomorphology,	palpation, percussion,	sequence of	
			clinical aspect,	auscultation);	diagnostic	
			differential	etiology,	measures incl.	
			diagnosis, clinical	pathogenesis and	modern digital	
			features,	pathogenesis and pathomorphology,	technologies	
			complications and	clinical aspect,		
			outcomes of diseases	differential		
			of internal organs;	diagnosis, clinical		
			patterns of	features,		
			functioning of a	complications and		
			healthy human body	outcomes of		
			and mechanisms for	diseases of		
			ensuring health from	internal organs;		
			the standpoint of the	patterns of		
			theory of functional	functioning of a		
			systems; features of	healthy human		
			the regulation of the	body and		
			functional systems of	mechanisms for		
			the human body in	ensuring health		
			pathological	from the		
			processes; methods	standpoint of the		
			of laboratory and	theory of		
			or incontroly and	01		

			instrumental	functional		
			examinations for	systems; features		
			assessing the state of	of the regulation		
			health, medical	of the functional		
			indications for	systems of the		
			conducting	human body in		
			investigations, rules	pathological		
			for interpreting their	processes;		
			results.	methods of		
			IPC-5.2. Be able to:	laboratory and		
			collect complaints,	instrumental		
			anamnesis of life and	examinations for		
			disease of the patient	assessing the state		
			and analyze the	of health, medical		
			information received;	indications for		
				conducting		
			conduct a complete	0		
			physical examination	investigations,		
			of the patient	rules for		
			(examination,	interpreting their		
			palpation,	results		
			percussion,			
			auscultation) and			
			interpret its results;			
			determine the			
			sequence of volume,			
			content and sequence			
			of diagnostic			
			measures incl.			
			modern digital			
			technologies			
6	PC-6	Able to send a	IPC-6.1. Knows:	general issues of	justify the	
		patient to	general issues of	organizing	need and	
		laboratory,	organizing medical	medical care for	scope of	
		instrumental	care for the	the population,	laboratory	
		examination, to	population, methods	methods of	examination of	
		a consultation	of laboratory and	laboratory and	the patient;	
		with specialist	instrumental	instrumental	justify the	
		doctors if there	examinations to	examinations to	need and	
		are	assess the state of	assess the state of	scope of	
		medical	health, medical	health, medical	instrumental	
		indications in	indications for	indications for	examination of	
		accordance with	conducting	conducting	the patient;	
		the current	examinations, rules	examinations,	justify the	
		procedures for	for interpreting their	rules for	need to refer	
		the provision of	results; procedures	interpreting their	the patient for	
		medical care,	for the provision of	results;	consultations	
		clinical	medical care, clinical	procedures for the	with specialist	
		recommendation	recommendations	provision of	doctors;	
		s	(treatment protocols)	medical care,	determine	
		s (treatment	on the provision of	clinical care,	medical	
		protocols) on		recommendations	indications for	
		* ·	medical care, standards of medical	(treatment		
		providing			the provision	
		medical care	care.	protocols) on the	of emergency,	
		taking into	IPC-6.2. Be able to:	provision of	including	
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		account the	justify the need and	medical care,	emergency	
		account the standards of medical care,	justify the need and scope of laboratory examination of the	standards of medical care	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 recommendation s (treatment protocols) on the medical care delivery taking into account the standards of medical care	need to refer the patient for consultations with specialist doctors; determine medical indications for the			
7 PC-7	Able to: make a differential diagnosis with other diseases/conditi ons including emergencies, establish a diagnosis taking	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	

ir	nformation about the	of the p	atient
p	oatient's disease;	by me	edical
ir	nterpret the data	specialists	; to
0	btained during the	carry	out
18	aboratory	differentia	1
ez	examination of the	diagnostic	es of
p	batient; interpret the	diseases	of
d	lata obtained during	internal o	rgans
th	he instrumental	from	other
ez	examination of the	diseases	
p	batient; interpret the		
d	lata obtained during		
C	consultations of the		
p:	batient by medical		
sj	pecialists; to carry		
01	out differential		
d	liagnostics of		
d	liseases of internal		
0	organs from other		
d	liseases		

4. Volume of the academic discipline and types of academic work

Type of educational work	Labor	intensity	Labor intensity (AH) in	
	volume in	volume in	seme	esters
	credit units	academic	5^{th}	6 th
	(CU)	hours (AH)	semester	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
TOTAL LABOR INTENSITY	1,0	36		36

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

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

	№	Compete nce code	Section name of the discipline	The content of the section in teaching units
ſ		UC-1	Introduction to	Medical genetics as a science. Clinical genetics, tasks. General
		GPC-5	clinical genetics.	patterns of pathogenesis of hereditary diseases. Classification
	1.	GPC-7	Personalized	and modern diagnosis of hereditary diseases. Hereditary
	1.	GPC-10	diagnosis and	pathology and methods of its study. genealogical method. The
		PC-5	treatment of	concept of personalized medicine. Personalized diagnosis and
		PC-6	hereditary	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.