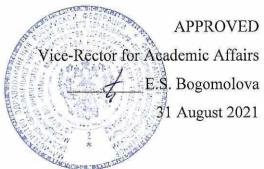
Federal State Budgetary Educational Institution of Higher Education "Privolzhsky Research Medical University" Ministry of Health of the Russian Federation



WORKING PROGRAM

Name of the academic discipline: BASICS OF CLINICAL GENETICS

Specialty: 31.05.01 GENERAL MEDICINE

Qualification: GENERAL PRACTITIONER

Department: **HOSPITAL PEDIATRICS**

Mode of study: FULL-TIME

Labor intensity of the academic discipline: 36 academic hours

The working program has been developed in accordance with the Federal State Educational Standard for the specialty 31.05.01 general medicine, approved by Order of the Ministry of Science and Higher Education of the Russian Federation No.965 of August 12, 2020.

Developers of the working program:

Khaletskaya O.V., MD, professor, head of the department of hospital pediatrics Yatsyshina E.E., PhD, associate professor of the department of hospital pediatrics Tush E.V., PhD, associate professor of the department of hospital pediatrics

The program was reviewed and approved at the department meeting (protocol No.4, April 8, 2021)

Head of the Department of hospital pediatrics, MD, professor

April 8, 2021

AGREED

Deputy Head of EMA ph.d. of biology _

Lovtsova L.V.

(Khaletskaya O.V.)

(signature)

April 8, 2021

- **1.** The purpose and objectives of mastering the academic discipline `Basics of clinical genetics` (hereinafter the discipline):
- 1.1. 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
- 1.2. Tasks of the discipline. Requirements to the deliverables of mastering the discipline As a result of completing the discipline, the student should

Know:

- 1. Fundamentals of ethics and deontology of medical practice, taking into account social, ethnic, confessional and cultural differences
- 2. Fundamentals and requirements for maintaining medical records in accordance with the legislation of the Russian Federation
- 3. Etiology, pathogenesis, morphogenesis, clinical picture, features of the course of diseases, classification of diseases
 - 4. Modern methods of clinical, laboratory and instrumental diagnosis of diseases.
 - 5. Principles of therapy for the most common diseases.
- 6. Principles and methods of providing medical care in various emergency conditions, indications for hospitalization.
- 7. Assessment of the quality of medical care using the main medical and statistical indicators
- 8. Theoretical foundations of computer science, collection, storage, processing, transformation, dissemination of information in medical and biological systems, the use of information computer systems in medicine and healthcare. Fundamentals of evidence-based medicine.

Be able to:

- 1. Use ethical and deontological principles in the work of a doctor, taking into account social, ethnic, confessional and cultural differences.
- 2. To issue medical documentation of various nature in medical organizations of various profiles.
- 3. Substantiate the principles of therapy for the most common diseases; prescribe rational therapy, apply drugs, their combinations for the treatment of children, write out prescriptions for medicines.
- 4. Collect anamnesis, conduct a physical examination of a patient of different ages; interpret the results of the examination, substantiate the diagnosis, determine the plan of examination and treatment.
- 5. Fill out medical documentation of various types in medical organizations of various profiles
 - 6. Identify life-threatening conditions and provide emergency care.
- 7. Analyze and evaluate the quality of medical care, the state of health of the population using the main medical and statistical indicators.
- 8. Use educational, scientific, popular science literature, the Internet for professional activities, methods of statistical processing of the material.

Possess

- 1. Principles of ethics and deontology of a doctor in clinical practice.
- 2. Registration of a medical history as a legal document and other documentation in medical organizations.
- 3. Algorithm for prescribing therapy; modern orders, standards and protocols for the provision of medical care.
- 4. Collecting anamnesis, methods of general clinical examination of patients, interpreting the results of laboratory, instrumental diagnostic methods, making a clinical diagnosis.
- 5. Algorithm and protocols for the implementation of basic medical diagnostic and therapeutic measures, the appointment of therapy.

- 6. Algorithm and protocols for the provision of medical care in urgent and life-threatening conditions.
 - 7. Algorithm for assessing the quality of medical care provided.
- 8. An algorithm for evaluating the main medical diagnostic and therapeutic measures to provide assistance to patients based on evidence-based medicine; presenting the results of the analysis for public discussion.
- 2. Position of the academic discipline in the structure of the General Educational Program of Higher Education (GEP HE) of the organization.
- **2.1.** The discipline **`Basics of clinical genetics`** refers to the core part of Block 1 of GEP HE 31.05.01 general medicine.

The discipline is taught in the 6^{th} semester/ 3^{rd} year of study.

- **2.2.** The following knowledge, skills and abilities formed by previous academic disciplines are required for mastering the discipline: anatomy, normal physiology, histology, biochemistry, propaedeutics of internal diseases
- **2.3.** Mastering the discipline is required for forming the following knowledge, skills and abilities for subsequent academic disciplines: faculty therapy, occupational diseases; hospital therapy, endocrinology; polyclinic therapy; as well as industrial practices: "Physician's assistant", "Physician's assistant of an outpatient clinic"

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

	Compet	The content of the	Code and name of the	As a result of master	ring the discipline, t	the students should:
№	ence code	competence (or its part)	competence acquisition metric	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; developing an action strategy to solve professional problems	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
2.	GPC-5	Able to assess morphofunctional,	IGPC-5.1. Knows: anatomy, histology,	anatomy, histology, embryology,	evaluate the basic	Has practical experience in:

	physiological conditions and pathological processes in the human body to solve professional problems	embryology, topographic anatomy, physiology, pathological anatomy and physiology of human organs and systems. IGPC-5.2. Be able to: evaluate the basic morphological and functional data, physiological conditions and pathological processes in the human body 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	topographic anatomy, physiology, pathological anatomy and physiology of human organs and systems	morphological and functional data, physiological conditions and pathological processes in the human body	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 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	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 unforeseen; features of medical care in emergency forms	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	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; 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

effects, adverse reactions, including unforeseen ones, resulting from diagnostic or therapeutic manipulations, the use of drugs and (or) medical devices, nondrug 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 medical of care: provision of emergency urgent medical care to patients with the most common diseases in accordance with the current procedures 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 nondrug treatment for patients with the most common diseases in accordance with the

procedures

for

treatment of the common most diseases of children and adults in accordance with current procedures for providing medical clinical care, recommendations, taking into account the standards of medical care; evaluation of the effectiveness and safety of the use of drugs, medical devices and nondrug 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 account the standards medical care; prevention and treatment of 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; providing medical care to patients with sudden acute diseases, conditions, exacerbation of chronic diseases without obvious signs of a threat to the patient's life in emergency form; the use of

medicines

and

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			providing medical care,			medical devices in
			clinical guidelines,			the provision of
			taking into account the			medical care in an
			standards of medical			emergency form
			care; prevention and			
			treatment of			
			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;			
			providing medical care			
			to patients with sudden			
			acute diseases, conditions,			
			exacerbation of 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			
			emergency form.			
4	GPC-	Able to	IGPC-10.1. Knows: the	the capabilities of	Apply modern	Has practical
1						
Ι.	10	understand the			information and	1
	10	understand the principles of	capabilities of reference	reference		experience in the use of modern
	10		capabilities of reference information systems	reference information systems	information and communication	experience in the
•	10	principles of	capabilities of reference information systems	reference	information and	experience in the use of modern
	10	principles of modern	capabilities of reference information systems and professional databases; methods of	reference information systems and professional	information and communication technologies to	experience in the use of modern information and
•	10	principles of modern information	capabilities of reference information systems and professional databases; methods of information retrieval,	reference information systems and professional databases; methods	information and communication technologies to solve the tasks of professional	experience in the use of modern information and bibliographic resources, the use
	10	principles of modern information technologies and	capabilities of reference information systems and professional databases; methods of information retrieval, information and	reference information systems and professional databases; methods of information	information and communication technologies to solve the tasks	experience in the use of modern information and bibliographic resources, the use
	10	principles of modern information technologies and use them to solve	capabilities of reference information systems and professional databases; methods of information retrieval, information and	reference information systems and professional databases; methods of information retrieval,	information and communication technologies to solve the tasks of professional activity; carry	experience in the use of modern information and bibliographic resources, the use of special software
	10	principles of modern information technologies and use them to solve the tasks of	capabilities of reference information systems and professional databases; methods of information retrieval, information and communication	reference information systems and professional databases; methods of information retrieval, information and communication technologies;	information and communication technologies to solve the tasks of professional activity; carry out an effective	experience in the use of modern information and bibliographic resources, the use of special software and automated
	10	principles of modern information technologies and use them to solve the tasks of professional	capabilities of reference information systems and professional databases; methods of information retrieval, information and communication technologies; modern medical and biological terminology;	reference information systems and professional databases; methods of information retrieval, information and communication technologies; modern medical and	information and communication technologies to solve the tasks of professional activity; carry out an effective search for information necessary to	experience in the use of modern information and bibliographic resources, the use of special software and automated information systems to solve standard tasks of
	10	principles of modern information technologies and use them to solve the tasks of professional	capabilities of reference information systems and professional databases; methods of information retrieval, information and communication technologies; modern medical and biological terminology; fundamentals of	reference information systems and professional databases; methods of information retrieval, information and communication technologies; modern medical and biological	information and communication technologies to solve the tasks of professional activity; carry out an effective search for information necessary to solve the tasks	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
	10	principles of modern information technologies and use them to solve the tasks of professional	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	reference information systems and professional databases; methods of information retrieval, information and communication technologies; modern medical and biological terminology;	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	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
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			information and		security	
			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			
			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			
L			information security			
5	PC-5	Able to collect	IPC-5.1. Knows: the	the legislation of	collect	
		complaints,	legislation of the	the Russian	complaints,	
		anamnesis of the	Russian Federation in	Federation in the	anamnesis of	
		patient's life and	the field of health care,	field of health care,	life and disease	
		illness, conduct	regulations and other	regulations and	of the patient	
		a complete	documents that	other documents	and analyze the	
		physical	determine the activities	that determine the	information	
		examination of	of medical	activities of medical	received;	
		the patient	organizations and	organizations and	conduct a	
		(examination,	healthcare workers;	healthcare workers;	complete	
		palpation,	method of collecting	method of	physical	
		percussion,	complaints, anamnesis	collecting	examination of	
		auscultation),	of life and disease of	complaints,	the patient	
		formulate a	the patient; a technique	anamnesis of life	(examination,	
		preliminary	for a complete physical	and disease of the	palpation,	
		diagnosis and	examination of the	patient; a technique	percussion,	
		draw up a plan for	patient (examination,	for a complete	auscultation)	
		laboratory and	palpation, percussion,	physical	and interpret its	
		instrumental	auscultation); etiology,	examination of the	results;	
		examinations of	pathogenesis and	patient	determine the	
		the patient	pathomorphology,	(examination,	sequence of	
			clinical aspect,	palpation,	volume, content	
			differential diagnosis,	percussion,	and sequence of	
			clinical features,	auscultation);	diagnostic	
			complications and	etiology,	measures incl.	
			outcomes of diseases of	pathogenesis and	modern digital	
			internal organs;	pathomorphology,	technologies	
			patterns of functioning	clinical aspect,		
			of a healthy human	differential		
			body and mechanisms	diagnosis, clinical		
			for ensuring health	features,		
			from the standpoint of	complications and		
			the theory of functional	outcomes of		
			systems; features of the	diseases of internal		
			regulation of the	organs; patterns of		
			functional systems of	functioning of a		
			the human body in	healthy human		
			pathological processes;	body and		
			methods of laboratory	mechanisms for		
1			and instrumental	ensuring health		
		İ	examinations for	from the standpoint		
			assessing the state of	of the theory of		

			health, medical	functional systems;		
			indications for	features of the		
			conducting	regulation of the		
			investigations, rules for	functional systems		
			interpreting their	of the human body		
			results.	in pathological		
			IPC-5.2. Be able to:	processes; methods		
			collect complaints,	of laboratory and		
			anamnesis of life and	instrumental		
			disease of the patient	examinations for assessing the state		
			and analyze the information received;	of health, medical		
			conduct a complete	indications for		
			physical examination	conducting		
			of the patient	investigations, rules		
			(examination,	for interpreting		
			palpation, percussion,	their results		
			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 need	
		patient to	general issues of	organizing medical	and scope of	
		laboratory,	organizing medical	care for the	laboratory	
		instrumental	care for the population,	population,	examination of	
		examination, to a consultation with	methods of laboratory and instrumental	methods of laboratory and	the patient; justify the need	
		specialist doctors	examinations to assess	instrumental	and scope of	
		if there are	the state of health,	examinations to	instrumental	
		medical	medical indications for	assess the state of	examination of	
		indications in	conducting	health, medical	the patient;	
		accordance with	examinations, rules for	indications for	justify the need	
		the current	interpreting their	conducting	to refer the	
		procedures for the	results; procedures for	examinations, rules	patient for	
		provision of	the provision of	for interpreting		
		medical care,	medical care, clinical	their results;	with specialist	
		clinical	recommendations	procedures for the	doctors;	
		recommendations	(treatment protocols)	provision of	determine	
		(treatment	on the provision of	medical care,	medical	
		protocols) on	medical care, standards	clinical	indications for	
		providing medical	of medical care. IPC-6.2. Be able to:	recommendations	the provision of	
		care taking into account the	justify the need and	(treatment protocols) on the	emergency, including	
		standards of	scope of laboratory	provision of	emergency	
		medical care, and	examination of the	medical care,	specialized,	
		also refer the	patient; justify the need	standards of	medical care	
		patient for	and scope of	medical care		
		providing	instrumental			
		specialized	examination of the			
		medical care in an	patient; justify the need			
		inpatient setting	to refer the patient for			
		or in a day	consultations with			
		hospital if there	specialist doctors;			
		are medical	determine medical			
		indications in	indications for the			
		accordance with	provision of			
		the current procedures for the	emergency, including emergency specialized,			
		medical care	medical care			
		delivery, clinical	modicai care			
		activery, citilical				

	1	manamman dation -				1
		recommendations (treatment protocols) on the				
		medical care delivery taking				
		into account the				
		standards of				
		medical care				
7	PC-7	Able to: make a	IPC-7.1. Knows:	etiology,	analyze the	
		differential	etiology, pathogenesis	pathogenesis and	results of the	
		diagnosis with other	and pathomorphology, clinical aspect,	pathomorphology,	patient's	
		diseases/condition	clinical aspect, differential diagnosis,	clinical aspect, differential	examination, if necessary,	
		s including	clinical features,	diagnosis, clinical	justify and plan	
		emergencies,	complications and	features,	the scope of	
		establish a	outcomes of diseases of	complications and	additional	
		diagnosis taking	internal organs;	outcomes of	examinations;	
		into account the	methods of laboratory	diseases of internal	interpret the	
		current international	and instrumental examinations for	organs; methods of laboratory and	results of collecting	
		statistical	assessing the state of	instrumental	information	
		classification of	health, medical	examinations for	about the	
		diseases and	indications for	assessing the state	patient's disease;	
		related health	conducting	of health, medical	interpret the	
		problems (ICD)	examinations, rules for	indications for	data obtained during the	
			interpreting their results; ICD.	conducting examinations, rules	during the laboratory	
			IPC-6.2. Be able to:	for interpreting	examination of	
			analyze the results of	their results; ICD	the patient;	
			the patient's		interpret the	
			examination, if		data obtained	
			necessary, justify and plan the scope of		during the instrumental	
			additional		examination of	
			examinations; interpret		the patient;	
			the results of collecting		interpret the	
			information about the		data obtained	
			patient's disease;		during	
			interpret the data obtained during the		consultations of the patient by	
			laboratory examination		medical	
			of the patient; interpret		specialists; to	
			the data obtained		carry out	
			during the instrumental		differential	
			examination of the		diagnostics of	
			patient; interpret the data obtained during		diseases of internal organs	
			consultations of the		from other	
			patient by medical		diseases	
			specialists; to carry out			
			differential diagnostics			
			of diseases of internal			
			organs from other diseases			
			arbeabeb			

4. Sections of the academic discipline and competencies that are formed when mastering them

№	Competen ce 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 PC-7	Introduction to clinical genetics. Personalized diagnosis and treatment of hereditary diseases.	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 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.

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

Type of educational work		Labor intensity			Labor inten	sity (AH) in
	volume	volume in volume in		seme	esters	
	credit (CU)	units	academic (AH)	hours	5 th semester	6 th 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

6. Content of the academic discipline

6.1. Sections of the discipline and types of academic work

	0.1. Sections of the discipline a	ma types c	n academi	C WOIK				
$N_{\underline{0}}$	Name of the section of the			Types of ac	cademic w	ork* (in A	H)	
	academic discipline	L	LP	P	CP	S	SIW	total
1.	Introduction to clinical genetics. Personalized diagnosis and treatment of hereditary diseases.	2			6		4	12
2.	Modern genetic research methods. Medical genetic counseling.	2			5		5	12

3.	Chromosomal diseases.	2		5	5	12
	Prenatal diagnosis of					
	congenital and hereditary					
	diseases.					
	TOTAL	6		16	14	36

- * L lectures; LP laboratory practicum; P practicals; CP Clinical Practice; S seminars; SIW student's individual work.
 - 6.2. Thematic schedule of educational work types:
 - 6.2.1 Thematic schedule of lectures

No	Name of lecture topics	Volume in AH
		semester 6
1.	Basics of clinical genetics.	2
2.	Modern technologies for diagnosing hereditary diseases.	2
3.	Chromosomal diseases.	2
	TOTAL (total - AH)	6

6.2.2. The thematic plan of laboratory practicums – this type of classes is not stipulated in the curriculum.

6.2.3. Thematic plan of clinical practice

No	Name of the topics of clinical practice	Volume in AH
		semester 6
1.	Hereditary pathology and methods of its study. genealogical method. Personalized diagnosis and treatment of hereditary diseases.	6
2.	Modern genetic research methods. Medical genetic counseling.	5
3.	Chromosomal diseases. Prenatal diagnosis of congenital and hereditary diseases.	5
	TOTAL (total - AH)	16

6.2.4. Thematic plan of seminars – this type of classes is not stipulated in the curriculum.

6.2.5. Types and topics of student's individual work (SIW)

№	Types and topics of SIW	Volume in AH
		semester 6
1.	Preparation for clinical practical classes,	5
	homework, preparation for current control	
2.	Working with lecture material	2
3.	Working with electronic resources on the PRMU	3
	distance education portal	
4.	The study of material submitted for independent	2
	study (separate topics, paragraphs), work with	
	literary sources	
5.	Preparation for testing, online testing	2
	TOTAL (total - AH)	14

7. Types of assessment formats for ongoing monitoring and mid-term assessment

	7. Types of assessment formats for ongoing monitoring and mid-term assessment							
	Se			Name of section		Assessment formats		
№	me ster No.	Types of	control	of academic discipline	Competence codes	types	number of test questions	number of test task options
	6	Current	Control of mastering the topic	Introduction to clinical genetics. Personalized	UC-1; GPC-5; GPC-7; GPC- 10; PC-5; PC- 6; PC-7	interview	4	93
1.		monitor ing	Monitorin g the student's individual work	diagnosis and treatment of hereditary diseases.	GPC-10; PC-5; PC-6; PC-7	test	15	50
2.	6	Current monitor	Control of mastering the topic	Modern genetic research methods.	UC-1; GPC- 5; GPC-7; GPC-10; PC-5; PC-6; PC-7	interview	1	43
2.		ing	Monitorin g the student's individual work	Medical genetic counseling.	UC-1; GPC- 5; GPC-7; GPC-10; PC-5; PC-6; PC-7	test	30	50
3.	6	Current monitor	Control of mastering the topic	Chromosomal diseases. Prenatal diagnosis of	UC-1; GPC- 5; GPC-7; GPC-10; PC-5; PC-6; PC-7	interview	1	43
3.		ing	Monitorin g the student's individual work	congenital and hereditary diseases.	UC-1; GPC- 5; GPC-7; GPC-10; PC-5; PC-6; PC-7	test	30	50
2.	6	Mid- term assess ment	Exam/ Credit		UC-1; GPC- 5; GPC-7; GPC-10; PC-5; PC-6; PC-7	situational tasks	1	10

8. Educational, methodological and informational support for mastering the academic discipline (printed, electronic publications, the Internet and other network resources)

8.1. Key literature references

No	Name according to bibliographic requirements	Number of copies		
		at the department	in the library	
1.	Nelson textbook of Pediatrics. V. 1: Nelson textbook	-	14	
	of Pediatrics. – [S. I.] : Elsevier, 2020. – 21ed. –			
	LXXV, 1900 p.: ill. – ISBN 978-0-323-56890-6.			
2.	Nelson textbook of Pediatrics. V. 2: Nelson textbook	-	14	
	of Pediatrics. – [S. I.]: Elsevier, 2020. – 21ed. –			
	LXXV, 1926 p.: ill. – ISBN 978-0-323-56890-6.			
	Khaletskaya, O. V. Basics of clinical genetics : a	Electronic resource Electronic resource		
	training manual for students / O. V. Khaletskaya, E. V.			

	Tush, A. N. Kolchina; FSBEI HE «PRMU» MOH	
I	Russia. – N. Novgorod: Publishing House of	
I	Privolzhskiy Research Medical University, 2021. – 1	
d	файл (2.16 Mб). – ISBN 978-5-7032-1406-0.	

8.2. Further reading

No	Name according to bibliographic requirements	Number of copies	
		at the department	in the library
1.	General biology. Molecular genetics: Handbook for international students / E. S. Knyazeva, O. M. Moskovtseva, I. N. Kalashnikov, T. G. Scherbatyuk. – FSBEI HE «PRMU» MOH Russia. – N. Novgorod: Publishing House of Privolzhskiy Research Medical University, 2021. – 1 файл. – ISBN 978-5-7032-1421-3.	Electronic	e resource
2.	Motor system disorders in neurological clinic: the textbook / V. N. Grigorieva, I. G. Stelnikova, T. A. Sorokina, A. A. Melnikov; FSBEI HE PRMU MOH Russia. – N. Novgorod: Publishing House of Privolzhskiy Research Medical University, 2021. – 1 файл (4.72 Мб). – ISBN 978-5-7032-1407-7.	Electronic resource	
3.	Nelson essentials of pediatrics / K. J. Marcdante, R. M. Kliegman. – 8th ed. – Philadelphia : Elsevier, 2019. – XIII, 818 p. : ill. – ISBN 978-0-323-52735-4.	1	4

8.3. Electronic educational resources for teaching academic subjects

8.3.1. Internal Electronic Library System of the University (IELSU)

№	Name of the electronic resource	Brief description (content)	Access conditions	Number of users
	Internal electronic library system (IELSU)	Proceedings of the teaching staff of the Academy: textbooks and teaching aids, monographs, collections of scientific papers, scientific articles, dissertations, abstracts of dissertations, patents.	computer on the Internet, using an individual login and password [Electronic	Not limited

8.3.2. Electronic educational resources acquired by the University

№	Name of the electronic resource	Brief description (content)	Access conditions	Number of users
	Electronic database	Educational literature +	from any	General
	"Student Advisor"	additional materials (audio,	computer on the	Subscription
		video, interactive	Internet, using an	
		materials, test tasks) for	individual login	
		higher medical and	and password	
		pharmaceutical education.	[Electronic	
		Editions are structured by	resource] - Access	
		specialties and disciplines	mode:	
		in accordance with the	http://www.studm	
		current Federal State	edlib.ru/	
		Educational Standards of		

	TT:-1 Df:1		
	Higher Professional Education.		
Electronic library system "BookUp"	Educational and scientific medical literature of Russian publishing houses, incl. translations of foreign publications.	from any computer located on the Internet by login and password, from the computers of the academy. Subscribed editions are available for reading. [Electronic resource] - Access mode: http://www.books-up.ru/	General Subscription
"Bibliopoisk"	Integrated search service "single window" for electronic catalogs, ELS and full-text databases. The results of a single search in the demo version include documents from domestic and foreign electronic libraries and databases available to the university as part of a subscription, as well as from open access databases.	access to the demo version of the Bibliopoisk search engine: http://bibliosearch .ru/pimu.	General Subscription
Domestic electronic periodicals	Periodicals on medical topics and higher education	- from the computers of the academy on the platform of the electronic library eLIBRARY.RU -magazines publishing house "Mediasphere" - from the computers of the library or provided library at the request of the user [Electronic resource] - Access mode: https://elibrary.ru/	
International scientometric database "Web of Science Core Collection"	Web of Science covers materials on natural, technical, social, humanities; takes into account the mutual citation of publications developed	[Electronic resource] - Access to the resource at: http://apps.webof knowledge.com	Free access from university computers

and provided by Thomson Reuters; has built-in search, analysis and	
management of bibliographic information.	

8.3.3 Open access resources

	8.3.3 Open access resources						
№	Name of the electronic resource	Brief description (content)	Access conditions				
	Federal Electronic Medical Library	Includes electronic analogues of printed publications and original electronic publications that have no analogues recorded on other media (dissertations, abstracts, books, magazines, etc.). [Electronic resource] - Access mode: http://нэб.рф/	from any computer on the Internet				
	Scientific electronic library eLIBRARY.RU	The largest Russian information portal in the field of science, technology, medicine and education, containing abstracts and full texts of scientific articles and publications. [Electronic resource] - Access mode: https://elibrary.ru/	from any computer on the Internet				
	Scientific electronic library of open access CyberLeninka	Full texts of scientific articles with annotations published in scientific journals in Russia and neighboring countries. [Electronic resource] - Access mode: https://cyberleninka.ru/	from any computer on the Internet				
	Russian State Library	Abstracts for which there are copyright agreements with permission for their open publication [Electronic resource] - Access mode: http://www.rsl.ru/	from any computer on the Internet				
	Reference and legal system "Consultant Plus"	Federal and regional legislation, judicial practice, financial advice, legislative comments, etc. [Electronic resource] - Access mode: http://www.consultant.ru/	from any computer on the Internet				
	Official website of the Ministry of Health of the Russian Federation	National Clinical Guidelines [Electronic resource] - Access mode: cr.rosminzdrav.ru - Clinical recommendations	from any computer on the Internet				
	Modern materials and clinical guidelines for the diagnosis and treatment of diseases in children	National Clinical Guidelines [Electronic resource] - Access mode: https://www.pediatr- russia.ru	from any computer on the Internet				
	Russian Society of Medical Genetics	Modern materials and clinical guidelines for the diagnosis and treatment of hereditary diseases	from any computer on the Internet				

	[Electronic resource] - Access				
	mode: http://www.romg.org				
Official website with basic	Modern materials and clinical	from any	computer	on	the
information on orphan diseases	guidelines for the diagnosis and	l Internet			
	treatment of orphan diseases				
	[Electronic resource] - Access				
	mode: www.orpha.net				

9. Material and technical support for mastering an academic discipline

- 9.1. List of premises for classroom activities for the discipline
- 1. 3 study rooms
- 2. Conference room
- 3. Computer classroom
- 9.2. List of equipment for classroom activities for the discipline
- 1.Multimedia complex
- 2. Information stands on the medical and protective regimen in the Hospital, nutrition and hygiene of patients.
- 3. Medical documentation: medical history, outpatient card
- 4. Archival and educational case histories
- 5. Slides and multimedia presentations of lectures.
- 6. Boards

9.3. A set of licensed and freely distributed software, including domestic production

Ite m no.	Software Wtware	number of licenses	Type of software Thin Client Operating System	Manufacture r Kovalev Andrey Alexandrovic h	Number in the unified register of Russian software	2471/05-18 from 28.05.2018
2	MyOffice is Standard. A corporate user license for educational organizations, with no expiration date, with the right to receive updates for 1 year.	220	Office Application	LLC "NEW CLOUD TECHNOLO GIES"	283	without limitation, with the right to receive updates for 1 year.
3	LibreOffice		Office Application	The Document Foundation	Freely distributed software	
4	Windows 10 Education	700	Operating systems	Microsoft	Azure Dev Tools for Teaching Subscriptio n	
5	Yandex. Browser		Browser	«Yandex»	3722	

6	Subscription to				23618/HN100
	MS Office Pro				30 LLC
	for 170 PCs for				"Softline
	FGBOU VO				Trade" from
	"PIMU" of the				04.12.2020
	Ministry of		Office		
	Health of Russia	170	Application	Microsoft	

10. List of changes to the working program (to be filled out by the template)

Federal State Budgetary Educational Institution of Higher Education
"Privolzhsky Research Medical University"
Ministry of Health of the Russian Federation
(FSBEI HE "PRMU" of the Ministry of Health of Russia)

Department of *Name of the department*

CHANGE REGISTRATION SHEET

working program for the academic discipline *BASICS OF CLINICAL GENETICS*

Field of	study / specialty / scie	entific specialty:		
		(code, na	(code, name)	
Training	g profile:			
	(nam	e) - for master's degree programs		
Mode of	f study:			
	,	ıral		
Position	Number and name of the program section	Contents of the changes made	Effective date of the changes	Contributor's signature
1				
			,	

signature

/ Khaletskaya O.V.

print name

Approved at the department meeting Protocol No. 4 of April, 8 2021

of hospital pediatrics, MD, professor

department name, academic title

Head of the Department