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

APPROVED
Vice-Rector for Academic Affairs
_____ E.S. Bogomolova
31 August 2021

WORKING PROGRAM

Name of the academic discipline: **FUNDAMENTALS OF MEDICAL GENETICS**

Specialty: **31.05.01 GENERAL MEDICINE**
(*code, name)*
Qualification: **GENERAL PRACTITIONER**

Department: **BIOLOGY**

Mode of study: **FULL-TIME**

Labor intensity of the academic discipline: **36 academic hours**

Nizhny Novgorod
2021

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. 988 of August 12, 2020.

Developers of the working program:
Full name, academic degree, title, position.

Kalashnikov Ilya Nikolaevich, Head of the Biology Department PRMU, PhD
Makhova Maria Alexandrovna, associate professor of the Department of Biology, PhD.

The program was reviewed and approved at the department meeting (protocol No. 8, August 17, 2021)
Head of the Department,
PhD of Biology, associate professor
(Kalashnikov I.N.)
August 17, 2021

(signature)

AGREED
Deputy Head of EMA ph.d. of biology _____ Lovtsova L.V.

(signature)

August 17, 2021

1. The purpose and objectives of mastering the academic discipline MEDICAL GENETICS (hereinafter – the discipline):

1.1. The purpose of mastering the discipline: (*participation in forming the relevant competencies).*

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

1.3. Requirements to the deliverables of mastering the discipline

As a result of completing the discipline, the student should

Know:

- the main directions of modern genetics: genomics, proteomics, epigenetics, ethnogenomics, facmacogenomics, metagenomics, modern approaches to gene therapy.
- fundamentals of monogenic diseases and principles of their diagnosis using modern advances in genetics.
- symptoms of common hereditary syndromes and the main approaches to their diagnosis.
- mechanisms of genetic and epigenetic variability.
- about mitochondrial diseases and diseases of genomic imprinting.
- on the prevalence and significance of hereditary diseases.
- prognosis for life, work capacity and social adaptation in the main nosological forms of hereditary diseases.

Be able to:

- to implement ethical and deontological aspects of medical activity in dealing with patients suffering from hereditary diseases.
- to build a pedigree in patients with signs of hereditary disease.

Possess:

- skills of molecular genetic research used to diagnose hereditary diseases.

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 **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.

2.2. The following knowledge, skills and abilities formed by previous academic disciplines are required for mastering the discipline:

1. biology,
2. normal anatomy,
3. histology with cytology and embryology,
4. normal physiology.

2.3. Mastering the discipline is required for forming the following knowledge, skills and abilities for subsequent academic disciplines:

1. microbiology,
2. virology;
3. immunology,
4. epidemiology,
5. medical ecology,
6. general hygiene.

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 for information and solutions based on action, experiment and experience	- 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 a preliminary diagnosis - 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	- 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 human heredity (cytogenetic method, biochemical method, genealogical method, twin method; the principle of DNA sequencing) - interpret the most significant changes in the results of laboratory	- 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 methods and identification of those changes that require referral of the patient to a geneticist.

				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 - 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	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 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	

				medical and genetic counseling	the importance of lifestyle for the preservation of human health and plan your life activities based on knowledge about a healthy lifestyle	

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

№	Competence code	Section name of the discipline	The content of the section in teaching units
1.	UC-1.	1. Molecular genetics is the basis of medical genetics. Genetic passport. Epigenetics. Ethnogenomics.	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.
		2. Methods of studying human genetics. Monogenic, chromosomal and genomic diseases.	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.
		3. Congenital diseases and malformations.	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.
		4. Methods of diagnosis of	Methods of DNA diagnostics. Use in clinical practice. • analysis of restriction fragment length polymorphism

		hereditary human pathologies and medical and genetic counseling.	• 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. 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.

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

Type of educational work	Labor intensity		Labor intensity (AH) in semesters
	volume in credit units (CU)	volume in academic hours (AH)	
Classroom work, including	***0,67***	***24***	
Lectures (L)	**0,17**	**6**	**6**
Laboratory practicum (LP)*	-	-	-
Practicals (P)	**0,5**	**18**	**18**
Seminars (S)	-	-	-
Student's individual work (SIW)	**0,33**	**12**	**12**
Mid-term assessment	-	-	-
credit/exam *(specify the type)*	-	-	-
TOTAL LABOR INTENSITY	**1**	**36**	**36**

6. Content of the academic discipline

6.1. Sections of the discipline and types of academic work

№	Name of the section of the academic discipline	Types of academic work* (in AH)					
		L	LP	P	S	SIW	total
1	Molecular genetics is the basis of medical genetics. Genetic passport. Epigenetics. Ethnogenomics.	6				3	9
2	Methods of studying human genetics. Monogenic, chromosomal and genomic diseases.			7		3	10
3	Congenital diseases and malformations.			4		3	7
4	Methods of diagnosis of hereditary human pathologies and medical and genetic counseling.			7		3	10
	TOTAL	**6**		**18**		**12**	**36**

* - L – lectures; LP – laboratory practicum; P – practicals; S – seminars; SIW – student's individual work.

6.2. Thematic schedule of educational work types:

6.2.1 Thematic schedule of lectures

№	Name of lecture topics	Volume in AH
		4 semester
1	Introduction to medical genetics. Differentiation of medical genetics. The human genome as the scientific basis of predictive medicine. Genetic passport.	2
2	Genomics. Proteomics. Genetic polymorphism. Ethnogenomics. Genomics and the creation of new drugs. The concept of drug metabolic safety. Pharmacogenetics.	2
3	Epigenetics and human diseases. Epigenetic determinants in malignant neoplasms. Genomic imprinting is an epigenetic system of gene regulation.	2
	TOTAL (total - AH)	**6**

6.2.2. The thematic plan of laboratory practicums (*if this type of classes is stipulated in the curriculum*)

The curriculum does not provide.

6.2.3. Thematic plan of practicals

№	Name of laboratory practicums	Volume in AH
		4 semester
1.	Methods of studying human genetics. The main clinical-genetic and research methods. 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.	**3**
2.	Chromosomal analysis. Cytogenetic research methods in medical genetics. Standard karyotyping. Fluorescent in situ hybridization (FISH method). Multicolored FISH methods. Examples of application in clinical practice. Spectral karyotyping (Spectral Karyotyping = SKY). 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.	**4**
3.	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. Agenesis. Aplasia. Atresia and stenosis. Absence of separate parts of the organ. Congenital hypoplasia. Congenital hypertrophy. Heteroplasia. Heterotopia. Ectopia. Dysgraphy or agraphy. Dyschromia. Macrosomy is the non-separation of organs or monozygotic twins. Persistence. Doubling of the organ. Additional organs. Small developmental anomalies.	**4**
4.	Methods of DNA diagnostics. Use in clinical practice. • analysis of restriction fragment length polymorphism • analysis of polymorphism of mini- and microsatellite sequences	**3**

	• 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 • Full genome sequencing (WGS) technology Applied aspects of the application of methods of molecular genetics and DNA diagnostics in clinical medicine.	
5.	Medical and genetic counseling as a type of specialized medical care for the population. 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. Family screening of hereditary pathology. Pre-symptomatic diagnosis (Huntington's disease, breast cancer, colon cancer)	**4**
	TOTAL (total - AH)	**18**

6.2.4. Thematic plan of seminars (*if this type of classes is stipulated in the curriculum)*
The curriculum does not provide.

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

№	Types and topics of SIW	Volume in AH
		4 semester
	– work with lecture material, providing for the study of lecture notes and educational literature;	**2**
	– completing homework for the lesson;	**2**
	– doing homework (problem solving, on-line testing);	**2**
	– study of the material submitted for independent study (separate topics);	**2**
	– preparation for the test	**4**
	TOTAL (total - AH)	**12**

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

№	Semester No.	Types of control	Name of section of academic discipline	Competence codes	Assessment formats		
					types	number of test questions	number of test task options
1.	4	Control of mastering the topic and monitoring the student's individual work	Molecular genetics is the basis of medical genetics. Genetic passport. Epigenetics. Ethnogenomics.	UC-1	On-line tests	-	30
2.	4	Control of mastering the topic and monitoring the student's individual work	Methods of studying human genetics. Monogenic,	UC-1	On-line tests Case-task Multi -level tasks	32	50

			chromosomal and genomic diseases.		and tasks		
3.	4	Control of mastering the topic and monitoring the student's individual work	Congenital diseases and malformations .	UC-1	On-line tests Multi -level tasks and tasks	- 2	50
4.	4	Control of mastering the topic and monitoring the student's individual work	Medical and genetic counseling as a type of specialized medical care for the population	UC-1	On-line tests Case-task Multi -level tasks and tasks	- 1 2	50
5.	4	Exam/ Credit	All sections of the discipline		Case-tasks	30	-
					Tasks	30	-
					On-line tests	-	300

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

№	Name according to bibliographic requirements	Number of copies	
		at the department	in the library
1.	Medical genetics / M. J. Bamshad, J. C. Carey, L. B. Jorde, R. L. White ; Jorde L. B. ; Carey J. C. ; Bamshad M. J. ; White R. L. – 3rd ed. – St. Louis : Mosby, 2006. – 363 с. : ил. мяг. – ISBN 978-0-323-04035-8.	-	49
2.	Connor, J. M. Essential medical genetics / J. M. Connor, M. A. Ferguson-Smith ; Connor, J. M. ; Ferguson-Smith, M. A. – 5th ed. – S.I. : Blackwell Science, 1998. – X, 236 p. : ill. – ISBN 9780865426665.	-	1

8.2. Further reading

№	Name according to bibliographic requirements	Number of copies	
		at the department	in the library
1	Бекиш, В. Я. Medical biology and general genetics: educational and methodical manual for foreign students of institutions of higher education in the specialty "dentistry" / V. Ya. Bekish, V. V. Bekish ; V. Ya. Bekish, V. V. Bekish. – Vitebsk : VSMU, 2021. – 274 p. – ISBN 9789855800591.	-	1
2	Gelehrter, T. D. Principles of medical genetics / T. D. Gelehrter, F. S. Collins, D. Ginsburg ; Gelehrter Thomas D. ; Collins,	-	4

	Francis S. ; Ginsburg David. – 2nd ed. – WILLIAMS & WILKINS, 1998. – 410 c. – ISBN 0-683-03445-6.		

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 (EBS) of PIMU	The works of the staff of the ADMU (textbooks, manuals, collections of tasks, manuals, laboratory work, monographs, etc.)	Access by individual login and password from any computer and mobile device	Not limited

8.3.2. Electronic educational resources acquired by the University and Open access resources

http://nbk.pimunn.net/MegaPro/Web

9. Material and technical support for mastering an academic discipline

9.1. List of premises for classroom activities for the discipline

1. Study rooms equipped with computers with Internet access, cabinets for storing microscopic equipment, cabinets for storing micro- and macro-preparations, study tables, laboratory equipment and equipment.

2. Lecture hall.

9.2. List of equipment for classroom activities for the discipline

1. Technical equipment: multimedia complexes (PC or laptop, projector, screen, presenters), interactive whiteboard.

Sets of slides, tables, diagrams, multimedia visual materials on various sections of the discipline. Micro- and macro-preparations, dummies. Situational tasks, test tasks on the studied topics,

- computer presentations on all topics of lecture and practical courses,

- educational videos by sections: cytogenetics, medical genetics, methods of DNA analysis.

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

Item no.	**Software**	**number of licenses**	**Type of software**	**Manufacturer**	**Number in the unified register of Russian software**	**Contract No. and date**
1	Wtware	100	Thin Client Operating System	Kovalev Andrey Alexandrovich	1960	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 TECHNOLOGIES"	283	without limitation, with the right to receive updates for 1 year.
3	LibreOffice		Office Application	The Document	Freely distributed	

				Foundation	software	
4	Windows 10 Education	700	Operating systems	Microsoft	Azure Dev Tools for Teaching Subscription	
5	Yandex. Browser		Browser	«Yandex»	3722	
6	Subscription to MS Office Pro for 170 PCs for FGBOU VO "PIMU" of the Ministry of Health of Russia	170	Office Application	Microsoft		23618/HN10030 LLC "Softline Trade" from 04.12.2020

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
NAME OF THE ACADEMIC DISCIPLINE

Field of study / specialty / scientific specialty: _____

(code, name)

Training profile: _____

(*name) - for master's degree programs*

Mode of study: _____

full-time/mixed attendance mode/extramural

Position	Number and name of the program section	Contents of the changes made	Effective date of the changes	Contributor's signature
1				

Approved at the department meeting
Protocol No. _____of _____20___

Head of the Department

_____ _____/_____

department name, academic title | signature | print name