


**FEDERAL STATE BUDGETARY
EDUCATIONAL INSTITUTION OF HIGHER EDUCATION
«AMUR STATE MEDICAL ACADEMY»
MINISTRY OF HEALTH OF THE RUSSIAN FEDERATION**

AGREED

Vice-Rector for Academic Affairs,

 N.V. Loskutova

April 17, 2025

Decision of the CCMC

April 17, 2025

Protocol No. 7


APPROVED

by decision of the Academic Council of the FSBEI
HE Amur SMA of the Ministry of Health of the
Russian Federation

April 22, 2025

Protocol No. 15

Acting Rector of the FSBEI HE Amur SMA of the
Ministry of Health of the Russian Federation

 I.V. Zhukovets

April 22, 2025



EDUCATIONAL PROGRAM

Discipline «Medical Genetics»

Specialty: 31.05.01 General Medicine

Course: 3

Semester: 6

Total hours: 108 hrs.

Total credits: 3 credit units

Control form: credit-test, 6 semester

Blagoveshchensk, 2025

The educational program of the discipline is designed in accordance with the requirements of the Federal State Educational Standard of Higher Education - specialist in specialty 31.05.01 General Medicine, approved by the order of the Ministry of Education and Science of Russia dated 08.12.2020 No. 988 (registered with the Ministry of Justice of Russia on 08.26.2020 No. 59493), BPEP HE (2021).

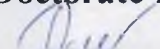
Authors:

Head of the Department of Pediatric Diseases, Holder of an Advanced Doctorate in Medical Sciences, Professor, E.B. Romantsova
Assistant of the Department of Pediatric Diseases, Ph.D. of Medical Sciences, O.S.Yutkina

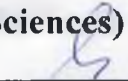
Reviewers:

Head of the Department of Pediatrics of the FSBEI HE Amur SMA, Ph.D. of Medical Sciences, Associate Professor, V.V. Shamraeva
Chief Physician of the State Autonomous Healthcare Institution of the Amur Region «Children's City Clinical Hospital», I.N. Molchanova


APPROVED at a meeting of the Department of Pediatric Diseases,
Protocol No. 8 dated March 26, 2025


Head of the Department, Holder of an Advanced Doctorate in Medical Sciences,
Professor  E.B. Romantsova

Conclusion of the Expert Commission on the review of the Educational Programs:
Protocol No. 1 dated April 16, 2025

Expert of the expert commission,
Holder of the Advanced Doctorate (Doctor of Sciences) in Medical Sciences,
Associate Professor  E.E. Molchanova

APPROVED at the meeting of the Cycle Methodical Committee No. 3:
Protocol No. 6 dated April 17, 2025

Chairman of the CMC No. 3
Holder of the Advanced Doctorate (Doctor of Sciences) in Medical Sciences,
Professor  V.V. Voitsehovsky

AGREED: Dean of the Faculty of Medicine,
Ph.D. of Medical Sciences, Associate Professor  N.G. Brush

April 17, 2025

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1. EXPLANATORY NOTE

1.1 Characteristics of the discipline

Medical genetics, as an independent scientific branch, has currently moved to one of the central places among medical and biological disciplines.

The need for genetic knowledge for a modern doctor is determined by the constant increase in the proportion of hereditary pathology in the structure of morbidity, mortality and disability of the population; the widespread use of genetic methodology, cytogenetic, molecular genetic methods to understand the molecular basis of the fundamental pathophysiological processes of hereditary diseases; the use of molecular genetic methods to decipher the interaction of heredity and environmental factors in the occurrence of common human pathology (atherosclerosis, coronary heart disease, diabetes mellitus, bronchial asthma, mental and oncological diseases, infectious diseases); the use of molecular genetic technologies to obtain more effective and less hazardous to health drugs; prospects for the widespread use of gene therapy to treat a number of diseases (the so-called "treatment" of genes and treatment with genes). This position is the basis for teaching and studying medical genetics as a clinical and preventive discipline. Since heredity and variability are integral properties of life, the study of genetics should be part of the theoretical and clinical training of a doctor

During the clinical training of students, medical genetics is studied as a clinical discipline. The general concept of teaching medical genetics in medical schools is to integrate genetic knowledge into the clinical thinking of the future doctor.

Classes in medical genetics are held in the 6th semester of the 3rd year: 15 practical classes and 22 hours of lectures. In the 7th semester, a test (midterm assessment) is held.

1.2 Objectives and tasks of the discipline

The purpose of teaching the discipline is to deepen basic knowledge and form systemic knowledge about the nature of hereditary human diseases, the causes of wide clinical polymorphism of pathology, diagnostics of genetic diseases using modern molecular genetic, cytogenetic research methods, prevention, principles of treatment of hereditary diseases.

Learning objectives of the discipline:

1. To teach students the skills of examining patients and their relatives in order to identify congenital and hereditary pathologies, to master the clinical features of hereditary pathologies, to assess the diagnostic and prognostic value of detected symptoms and morphogenetic variants (microanomalies) of development.
2. To give students an idea of the nature of human hereditary diseases, their etiology, pathogenesis, clinical presentation, diagnostics, and treatment.
3. To teach students to master the clinical- genealogical method with the formation of a preliminary conclusion about the type of inheritance of pathology in a specific family.
4. To train students in approaches and methods for identifying individuals with an increased risk of developing widespread non-infectious diseases (multifactorial diseases).
5. To teach the basic methods of diagnosing the most common forms of hereditary pathology, including modern methods of cytogenetic, biochemical and molecular genetic diagnostics.
6. To provide an idea of the stages of implementation, methods of medical genetic counseling, prenatal diagnostics and screening programs .
7. To familiarize students with the moral and legal norms of providing medical and genetic care.
8. To familiarize students with computer diagnostic programs and principles of computer diagnostics of hereditary diseases,
9. Knowledge of the principles of interaction between the medical genetic service and all practical health services and indications for organizing the flow of patients .

1.3 Place of the discipline in the structure of the main professional educational program higher education.

In accordance with the Federal State Educational Standard of Higher Education - a specialist in specialty 31.05.01 General Medicine (2020), the discipline "Medical Genetics" belongs to Block 1 (B 1 .B.50) "Basic Part". The total workload is 3 ZE (108 hours).

1.4 Requirements for students

To study the discipline, knowledge, skills and abilities formed by previous disciplines are necessary:
Latin
Knowledge : Basic medical and pharmaceutical terminology in Latin.
Skills: be able to apply knowledge for communication and obtaining information from medical literature, medical documentation. (II - III level)
Skills : applies medical and pharmaceutical terminology in Latin in professional activities
Professional foreign language
Knowledge: Basic medical and pharmaceutical terminology in a foreign language. (II - III level)
Skills : be able to apply knowledge for communication and obtaining information from foreign sources.
Skills: applies medical and pharmaceutical terminology in a foreign language in professional activities
History of Medicine
Knowledge: outstanding figures in medicine and health care, Nobel laureates, outstanding medical discoveries in the field of medical genetics, the influence of humanistic ideas on medicine. (II - III level)
Skills: be able to competently and independently present and analyze the contribution of domestic and foreign scientists to the development of medical genetics.
Skills: the ability to competently conduct a scientific discussion on the most important issues of the general history of medicine
Bioethics
Knowledge : moral and ethical standards, rules and principles of professional medical conduct, rights of the patient and the doctor, basic ethical documents regulating the activities of the doctor. (II - III level)
Skills: be able to build and maintain working relationships with patients and other team members.
Skills: skills of argumentation, conducting discussions, resolving complex ethical and legal situations.
Anatomy
Knowledge: Age-related anatomical features of organs and systems of children and adolescents.
Skills: use knowledge of the anatomical features of a child's body at different age periods
Skills: apply terminology in the field of structure and topography of organs and tissues, organ systems and apparatuses of the human body.
Histology, embryology, cytology
Knowledge: Cell structure. Spermatogenesis, oogenesis, their stages. Critical periods for the formation of organs and systems in embryogenesis. (II - III level)
Skills: be able to determine and predict the impact of a teratogenic factor on fetal development depending on the duration of exposure.
Skills: use histo -functional characteristics of the main systems of the body, the patterns of their embryonic development, as well as functional, age-related and protective-adaptive changes in organs and their structural elements.

Biology
Knowledge: laws of genetics and its importance for medicine; patterns of heredity and variability in individual development as the basis for understanding the pathogenesis and etiology of hereditary and multifactorial diseases; biosphere and ecology (II - III level).
Skills: be able to analyze patterns of heredity and variability in the development of hereditary diseases, multifactorial pathology.
Skills: conduct comparative analysis, approaches to classification and methodology of differential diagnostics of hereditary and non-hereditary pathology
Microbiology, virology
Knowledge: The importance of microbial genetics in the development of general genetics. Microbiological diagnostics of infectious diseases. (Level II)
Skills: be able to analyze the results of diagnostics of infectious diseases;
Skills: application of methods of laboratory diagnostics of infectious diseases, interpretation of the results obtained during microbiological, molecular biological and immunological studies of biological fluids, virus-containing materials and pure cultures of microbes.
Physics, Mathematics. Medical informatics. Medical biophysics
Knowledge: mathematical methods for solving intellectual problems and their application in medicine; theoretical foundations of computer science, collection, storage, search, processing, transformation, distribution of information in medical and biological systems, use of information computer systems in medicine and health care; principles of operation and design of equipment used in medicine, principles of physical and mathematical laws reflected in medicine (II - III levels).
Skills: be able to use educational, scientific, popular science literature, the Internet for professional activities, work with equipment taking into account safety regulations .
Skills: works with computer technology to perform calculations using formulas, performs statistical processing of experimental results.
Bioorganic chemistry in medicine. Biochemistry
Knowledge : chemical and biological essence of processes occurring in a living organism at the molecular and cellular levels . Mechanisms of biochemical homeostasis, main indicators of metabolism in norm and pathology, modern methods of biochemical research in the clinic . (II - III level).
Skills : be able to analyze the contribution of chemical and biochemical processes in the pathogenesis of hereditary pathology, interpret the results of the most common laboratory diagnostic methods to identify disorders in the metabolism of proteins, fats, carbohydrates, and microelements.
Skills: apply chemical and physicochemical methods of analysis in medicine
Pathophysiology, clinical pathophysiology
Knowledge: Typical forms of metabolic disorders of proteins, carbohydrates, lipids, nucleic acids, minerals, pathophysiology of tumor growth, allergies.
Skills : be able to analyze the significance of disturbances in the regulation of the metabolism of proteins, carbohydrates, lipids, nucleic acids, and minerals in the formation of phenotypic traits.
Skills: conducting pathophysiological analysis of data on pathological syndromes, pathological processes, conditions and reactions, forms of pathology and individual diseases.
Pathological anatomy, clinical pathological anatomy
Knowledge: Pathological processes of hereditary and non-hereditary diseases . pathological anatomy of congenital malformations.
Skills: Be able to analyze the significance of teratogenic factors for the formation of congenital malformations.
Skills: clinical and anatomical analysis based on a comparison of morphological and clinical manifestations of diseases at all stages of their development.
Propaedeutics of internal diseases

Knowledge: collection of complaints, anamnesis of life and disease, physical examination
Skills: be able to interpret complaints, life and disease history, physical examination data (II - III level)
Skills: application of methods of direct examination of the patient (questioning, examination, palpation, percussion, auscultation, measurement of blood pressure, examination of the properties of the arterial pulse, etc.).

1.5 Interdisciplinary links with subsequent disciplines

The knowledge and skills acquired in the discipline "Medical Genetics" are necessary for studying subsequent disciplines

No.	Name of subsequent disciplines	Medical genetics
1.	Dermatovenereology	+
2.	Hospital therapy	+
3.	Outpatient therapy	+
4.	Hospital surgery, pediatric surgery	+
5.	Obstetrics and gynecology	+
6.	Infectious diseases	+
7.	Oncology, radiation therapy	+
8.	Ophthalmology	+

1.6. Requirements for the results of mastering the discipline

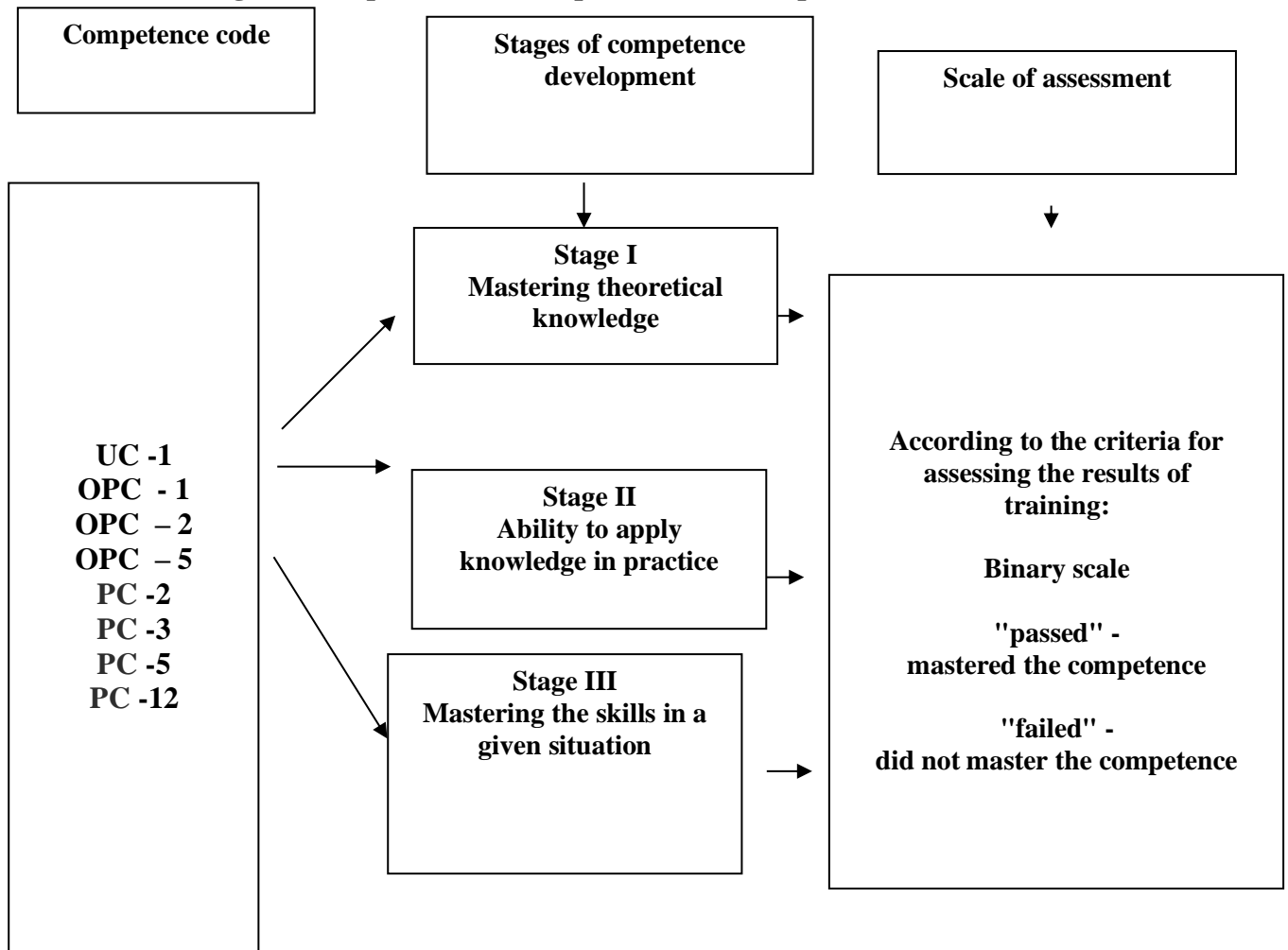
The study of the discipline "Medical Genetics" is aimed at the formation/improvement of the following competencies: universal (UK) - UK-1 (ID UK-1.1, 1.2, 1.5) ; general professional (OPK) – OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) and professional (PC) – PC-2 (ID PC 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (ID PC 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .

No. /p	Code and name of competence	Code and the name of the indicator of achievement of competence	As a result of studying the academic discipline, the student must:		
			Know	Be able to	To own
Universal competencies					
1	UK-1. Capable of carrying out a critical analysis of problematic situations based on a systems approach and developing an action strategy	ID UK-1.1. Analyzes the problem situation as a system, identifying its components and the connections between them. ID UK-1.2. Identifies gaps in information needed to solve problem situations and designs processes to eliminate them. ID UK-1.5. Critically evaluates the reliability of information sources, works with contradictory information from different sources.	The role of medical genetics in protecting children's health, interdisciplinary interactions for organizing high-quality care for children. Sources for reliable medical information.	Using an interdisciplinary approach, it is necessary to properly organize the interaction of the local pediatrician with specialists of different profiles. Receive information from trusted sources.	The skill of assigning "routing" to a patient with a hereditary pathology to establish a diagnosis and prescribe treatment, using reliable and proven sources of information .
General professional competencies					
2	OPK-1 Able to implement moral and legal norms, ethical and deontological principles in professional activities	ID OPK-1.1. Carries out professional activities in accordance with ethical standards and moral principles. ID OPK-1.2. Organizes professional activities, guided by legislation in the field of healthcare, knowledge of medical ethics and deontology.	Ethical and deontological aspects of the relationship "doctor-doctor", "doctor-patient with hereditary pathology " .	When working with patients with hereditary pathology, observe all principles of medical ethics and deontology.	Communication skills with patients with hereditary pathology, relatives, colleagues, applying ethical standards and moral principles.
3	OPK-2 Capable of conducting and monitoring the effectiveness of measures for prevention, healthy lifestyle promotion and sanitary and hygienic education .	ID OPK-2.2. Promotes a healthy lifestyle aimed at improving sanitary culture and preventing diseases of patients; organizes events on sanitary and hygienic education and the formation of healthy lifestyle skills. ID OPK-2.6. Assesses the characteristics of population health and environmental factors that affect the body, knows the biophysical mechanisms of such impact.	Questions of the etiology of the disease and factors contributing to the development of hereditary disease, with questions of prevention of identified deviations.	To draw up a plan for the dispensary observation of a patient with a hereditary pathology.	Skills in organizing events on sanitary and hygienic education and the formation of healthy lifestyle skills for the purpose of preventing hereditary diseases .
4	OPK-5 Capable of assessing morphofunctional,	ID OPK-5.2. Knows the etiology, pathogenesis, morphogenesis, pathomorphosis of disease development, basic concepts of nosology.	Etiopathogenesis of various hereditary diseases, indicators of the morphofunctional and	Recognize the pathological process in the patient's body, use indicators of the	Skills in diagnosing various hereditary pathologies.

	physiological states and pathological processes in the human body to solve professional problems	<p>ID OPK-5.3. Knows the indicators of the morphofunctional, physiological state of a healthy person and can measure/determine them.</p> <p>ID OPK-5.4. Applies indicators morphofunctional, physiological state and pathological process for examination of the human body in order to establish a diagnosis, prescribe treatment and monitor its effectiveness and safety.</p>	physiological state of a healthy and sick patient.	morphofunctional, physiological state and pathological process to examine the body of a patient with hereditary pathology.	
Professional competencies					
5	<p>PC-2</p> <p>Able to collect and analyze complaints, life history and medical history of the patient in order to establish a diagnosis</p>	<p>ID PC-2 .1. Establishes contact with the patient.</p> <p>ID PC- 2.2 . Collects complaints, specifies them, highlighting the main and secondary ones.</p> <p>ID PC- 2.3. Collects and analyzes information about the onset of the disease, the presence of risk factors, the dynamics of the development of symptoms and the course of the disease.</p> <p>ID PC- 2.4 . Analyzes the timing of the first and repeated requests for medical care, the volume of therapy performed, and its effectiveness.</p> <p>ID PC -2.5 . Collects and evaluates life history, including past illnesses, injuries and surgeries, hereditary, professional, epidemiological history.</p>	Methods of collecting anamnesis and its assessment in patients with hereditary pathology. Main clinical manifestations (symptoms, syndromes) of hereditary diseases.	In patients with hereditary pathology, analyze complaints, life history, diseases, volume of therapy, data on past illnesses, injuries and surgeries, hereditary (genealogical), allergological and epidemiological history.	Skills in collecting and analyzing anamnestic data, compiling a family tree.
6	<p>PC-3</p> <p>Able to conduct a physical examination of a patient, analyze the results of additional examination methods in order to establish a diagnosis</p>	<p>ID PC-3.1 . Conducts a complete physical examination of the patient (inspection, palpation, percussion, auscultation) and interprets its results .</p> <p>ID PC-3.2. Justifies the necessity, volume, sequence of diagnostic measures (laboratory, instrumental) and referral of the patient to specialist doctors for consultations</p> <p>ID PC-3.3. Analyzes the results of the patient examination, if necessary, justifies and plans the scope of additional studies.</p> <p>ID PC-3.4. Interprets and analyzes the results of collecting information about the patient's disease, data obtained during laboratory and instrumental examinations and during</p>	Methods of physical examination of patients (inspection, palpation, percussion, auscultation) with hereditary pathology and assessment of its results. Methods of laboratory and instrumental diagnostics of hereditary diseases.	Examine a patient with a hereditary pathology (inspection, palpation, percussion, auscultation) To draw up an algorithm for diagnostic search, a plan for laboratory and instrumental examination of a patient with hereditary pathology . To highlight the leading clinical and laboratory syndromes To evaluate the differential diagnostic significance of syndromes characteristic of the diseases under study.	Skills in interpreting and analyzing the results of collecting information about the disease of a patient with hereditary pathology , data obtained during laboratory and instrumental examinations and during consultations with specialist doctors, skills in substantiating and planning the scope of additional research.

		consultations with specialist doctors, and, if necessary, justifies and plans the scope of additional research.		Conduct differential diagnosis of diseases with similar clinical symptoms Assess the severity of the disease.	
7	PC-5 Capable prescribe treatment to patients	ID PC-5. 2. Prescribes medications, medical devices and therapeutic nutrition taking into account the diagnosis, age and clinical picture of the disease in accordance with the current procedures for the provision of medical care, clinical recommendations, taking into account the standards of medical care ID PC-5. 3. Prescribes non-drug treatment taking into account the diagnosis, age and clinical picture of the disease in accordance with the current procedures for the provision of medical care, clinical recommendations, taking into account the standards of medical care.	Basic principles of treatment, including non-drug methods of hereditary diseases. Mechanism of action of prescribed drugs, indications and contraindications.	To draw up a treatment plan for patients with hereditary pathology , taking into account age, diagnosis and clinical picture, in accordance with current clinical guidelines (treatment protocols), procedures for providing medical care and taking into account standards of medical care. Prescribe diet therapy, drug and non-drug therapy.	The ability to treat hereditary pathology in patients of different ages.
8	PC-12 Ready to maintain medical records, including in electronic form.	ID PC-12.1. Fills out medical documentation, including in electronic form. ID PC-12.2. Works with personal data of patients and information constituting a medical secret.	Basic medical documents of outpatient and inpatient medical institutions.	Work with personal data of patients with hereditary pathology and information constituting a medical secret.	Skills in filling out medical forms documentation , including in electronic form.

1.7 Stages of competencies development and description of assessment scales



1.8 . Forms of training organization and types of control

Form of organization of students' training	Brief characteristic
Lectures	The lecture material contains key and most problematic issues of the discipline, which are most significant in the training of a specialist.
Practical classes	They are intended for the analysis (reinforcement) of theoretical principles and monitoring their assimilation with subsequent application of the acquired knowledge during the study of the topic.
Interactive forms of education	solving situational problems with subsequent discussion, clinical analysis of thematic patients, interactive survey, completing tasks, small group method, discussions, online course of the discipline in the Moodle system , testing in the Moodle system .
Participation in the department's research work, student circle and conferences	preparation of oral presentations, poster presentations for presentation at a student club or scientific conference, writing theses and abstracts on the chosen scientific field; preparation of a literature review using educational, scientific, reference literature and Internet sources.
Types of control	Brief description
Incoming inspection	Testing theoretical knowledge, skills and abilities formed by previous disciplines The entrance knowledge control includes: testing in the Moodle system (test of incoming knowledge control) The results of the incoming inspection are systematized, analyzed and used by the teaching staff of the department to develop measures to improve and update the teaching methods of the discipline.
Current control	Current knowledge control includes: checking the solution of situational problems and assignments completed independently (extracurricular independent work); assessment of the assimilation of theoretical material (oral survey); control over the supervision of thematic patients during practical classes and the preparation of medical documentation; testing in the Moodle system on all topics of the discipline (tests include questions of a theoretical and practical nature); individual assignments (practical and theoretical) for each topic of the discipline being studied.
Final control	The final assessment of knowledge includes: testing in the Moodle system for the entire discipline in the 6th semester.
Intermediate certification	The midterm assessment is presented as a test at the end of the 6th semester: it includes an assessment of knowledge of theoretical material (interview on tickets).

II . STRUCTURE AND CONTENT OF THE DISCIPLINE

2.1 Scope of the discipline and types of educational activities

Types of educational work	Total hours	Semester
		6
Lectures	22	22
Practical classes	50	50
Independent work of students	36	36
Total labor intensity in hours	108	108
Total workload in credit units	3	3

Note: The curriculum for the discipline "Medical Genetics" for students of the Faculty of Medicine includes theoretical (lecture course) and practical training (practical classes). The training is conducted during one semester (VI) and includes 22 hours of lectures, 50 hours of classroom practical training and 36 hours of extracurricular independent work . in the form of midterm assessment – credit in the 6th semester.

2.2 Thematic plan of lectures and their brief content

No.	Topic of lectures	Codes of formed competencies	Labor intensity (hours)
1.	Introduction to medical genetics. Basic principles and concepts of medical genetics. Brief history of medical genetics. Subject and tasks of medical genetics. Importance of medical genetic knowledge in the practical work of a physician. Place of medical genetics in the system of medical knowledge, relationship of medical genetics with other clinical and medical-preventive disciplines. Increase in the share of hereditary pathology in the structure of morbidity, mortality and disability of the population, male and female infertility. Population-genetic, ecological, socio-economic and demographic aspects of hereditary pathology. Main stages of development of human genetics. History of development of medical genetics. Directions of development of modern genetics, human genetics, medical and clinical genetics.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
2.	Etiology and pathogenesis of hereditary diseases. Genetic bases of homeostasis. Genetic control of pathological processes. Mutations as etiological factors. Classification of mutations. Causes of mutations. Spontaneous and induced mutagenesis (methods of studying, recording and monitoring mutagenic effects of anthropotechnical environmental factors). Features of the pathogenesis of hereditary diseases in connection with the nature of damage to genetic structures. Gene and phenotypic correlations in chromosomal and gene diseases. Pathogenesis of diseases with hereditary predisposition and risk factors.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
3.	Semiotics and principles of clinical diagnostics of hereditary pathology. Classification of hereditary diseases. Syndromological method, "portrait" diagnostics, "family" approach - the main principles of diagnostics of hereditary diseases. Clinical and morphological examination. Stigmas of dysembryogenesis . Stages of diagnosis of hereditary diseases. Signs, classification of hereditary pathology.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5	2

		PC-12	
4.	Methods of diagnostics of hereditary diseases . Clinical and genealogical method. Cytogenetic methods. Biochemical methods: presumptive diagnostics, confirmatory diagnostics. Modern methods: automated analysis of amino acids, liquid and gas chromatography, mass spectrometry , nuclear magnetic resonance, radioimmunochemical and immunoenzyme methods. Molecular genetic methods: characteristics of the main methodological techniques. PCR. Diagnostic methods: sequencing , analysis of conformational polymorphism of single-stranded DNA, etc. Gene linkage method.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
5.	Morphogenetic variants of development. Developmental defects. Morphogenetic variants of development, genesis, postnatal modification. General and specific morphogenetic variants: significance in diagnostics of hereditary syndromes and congenital conditions. Developmental defects: primary and secondary. Isolated, systemic and multiple. Etiological heterogeneity of congenital developmental defects. Concept of syndrome, association, deformation, dysplasia. Teratogenic termination period. Mutation process and environmental factors. Radiation mutagenesis.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
6.	Chromosomal syndromes. General characteristics of chromosomal syndromes. Etiology. Cytogenetics of chromosomal syndromes. Chromosomal imprinting. Pathogenesis of chromosomal syndromes. Mechanisms of developmental disorders and occurrence of developmental defects in chromosomal syndromes: change in gene dosage, disruption of developmental "canalization", "prohibited" pathways of morpho- , histo- , organogenesis. General clinical characteristics of chromosomal syndromes. Methods of diagnosing chromosomal syndromes. Outcomes of chromosomal syndromes. Possibilities of therapy and rehabilitation of patients.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
7.	Monogenic diseases. General characteristics of monogenic pathology. Prevalence of diseases in different populations, ethnic groups and representatives of different nationalities. General issues of etiology and pathogenesis of monogenic diseases. Types of gene mutations, mutations of mitochondrial genes. Mechanisms of pathogenesis of monogenic diseases. Concept of gene- , pheno- , and normocopies . Classification of monogenic diseases. Monogenic syndromes of multiple congenital malformations: general features, examples. Clinical genetics of individual forms of monogenic diseases. General principles of treatment of hereditary diseases, rehabilitation and social adaptation of patients.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
8	Hereditary metabolic diseases. Modern classification, brief characteristics of groups, difficulties of classification. Monogenic diseases with established damaged biochemical function; diseases with identified mutant gene product. Pathogenesis scheme of hereditary metabolic diseases, metabolic blocks. Clinical genetics of individual forms of metabolic diseases. General principles of treatment of metabolic diseases, rehabilitation and social adaptation of patients.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2

9	Diseases with hereditary predisposition. The concept of predisposition. Specific mechanisms of realization of hereditary predisposition. General characteristics of multifactorial diseases: high frequency in the population; continuous series of conditions from subclinical to pronounced clinical manifestations; the nature of age and gender differences; features of the spread of predisposition genes and the incidence of diseases in families. Risk of occupational diseases from a genetic point of view.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
10	Principles of treatment of hereditary diseases . Treatment of hereditary diseases. General principles and approaches to therapy of hereditary pathology. Symptomatic therapy. Pathogenetic treatment. Etiological therapy of hereditary diseases. Ways and methods (selection of vector and target tissues for gene therapy). Gene therapy is the introduction of genetic material (DNA or RNA) into a cell, the function of which it changes. Genetic engineering is a set of techniques, methods and technologies for obtaining recombinant RNA and DNA, isolating genes from the cells of an organism, manipulating genes and introducing them into the cells of other organisms.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
11	Prevention of hereditary pathology. Ethical issues of medical genetics. Types and directions of prevention of hereditary diseases: pregametic , prezygotic , prenatal and postnatal . Ways of carrying out preventive measures: penetrance and expressivity management; elimination of embryos and fetuses; family and childbirth planning; environmental protection. Forms of preventive measures: medical and genetic counseling; prenatal diagnostics; mass screening programs ; periconceptional prevention, "genetic" medical examination of the population (registers); environmental protection and control over the mutagenicity of environmental factors. Deontological and ethical issues of medical genetics.	UK-1 OPK-1 OPK-2 OPK-5 PC-2 PC-3 PC-5 PC-12	2
Total hours			22

2.3 Thematic plan of practical classes and their content

Practical classes in the discipline "Medical Genetics" are a mandatory section and represent a type of educational activity directly focused on the professional training of students. The main goal is the integration of genetic knowledge into the structure of clinical thinking of a general practitioner as a basis for diagnosis, prevention and treatment of diseases and strengthening the health of the population.

No.	Topics of practical classes	Contents of practical classes of the discipline	Codes of the formed competencies and indicators of their achievements	Types of control	Labor intensity (hours)
1	Introduction to Medical Genetics. Heredity and Pathology.	<p>Theoretical part: Definition of the subject. Objectives of medical genetics. History of the development of medical genetics. Interaction of heredity and environment in the formation of resistance and predisposition to diseases. Etiology of hereditary diseases. Classification of hereditary diseases. Mutations as an etiological factor. Pathogenesis of hereditary diseases (molecular, cellular, tissue levels). Determining and modifying influence of genetic constitution on the development of disease. Heredity and clinical picture. Heredity and disease outcomes. Contribution of mutations to intrauterine fetal death, perinatal and early childhood mortality.</p> <p>Practical part: Complete exercises and tasks according to the example, work with handouts, scientific, medical and reference literature. Conduct supervision of patients with hereditary pathology, draw up a pedigree chart in the workbook. Calculate the risk of hereditary pathology using the Punnett square . Recognize common manifestations of hereditary pathology. To determine the role of hereditary and environmental factors in the development of pathology.</p>	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5)	Current Testing Frontal survey Interactive survey	3.3
2	Semiotics of hereditary pathology, principles of clinical diagnostics. Clinical and morphological examination.	<p>Theoretical part: General and specific semiotics of hereditary pathology. Features of the semiotics of hereditary diseases. Syndromological method, "portrait" diagnostics, "family" approach - the main principles of diagnostics of hereditary diseases. Clinical and morphological examination. Stigmas of dysembryogenesis . Stages of diagnosis of hereditary diseases. Signs of hereditary diseases. Classification of hereditary pathology.</p> <p>Practical part: Examine the patient for hereditary pathology.</p>	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) ,	Current Testing Frontal survey Interactive survey	3.3

		Recognize common manifestations of hereditary pathology. Collect anamnestic data and genealogical information, compile a pedigree, present it in graphic form and analyze the inheritance of the disease or disease symptom in the family. Present the results of the clinical , genetic and laboratory examination in the form of diaries and conclusions in the patient's medical history.			
3	Methods of medical genetics. Screening programs. Neonatal screening.	<p>Theoretical part : Clinical and genealogical method. Definition and essence of the method. Stages of the clinical and genealogical examination. Basic concepts: pedigree, proband, pedigree legend, symbols. Criteria for different types of inheritance: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, mitochondrial . Genealogical analysis of multifactorial diseases. Cytogenetic methods. Definition, essence, indications for using the method. The concept of karyotype. Modern methods of chromosome research. The value of the cytogenetic method in clinical practice. Biochemical methods. The value of biochemical methods in the diagnosis of hereditary metabolic diseases and multifactorial diseases. Definition, essence, indications for using the method. Molecular genetic methods. Definition, essence, indications for using the method. Versatility of DNA diagnostic methods, possibilities of their use. Polymerase chain reaction. Resolving capabilities of molecular genetic methods in diagnostics of hereditary diseases. Prenatal, preclinical diagnostics and diagnostics of heterozygous states. Screening programs - preclinical diagnostics of hereditary diseases. Principles of selection of nosologies. Neonatal screening.</p> <p>Practical part: Examine the patient for hereditary pathology, recognize common manifestations of hereditary pathology, diagnose congenital morphogenetic variants, correctly use the appropriate terminology when describing the clinical picture (phenotype) of the patient. Collect anamnestic data and genealogical information, compile a pedigree, present it in graphic form and analyze the inheritance of a disease or a disease trait in a family. Formulate a presumptive diagnosis of hereditary pathology and some of the most common monogenic</p>	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	Current Testing Frontal survey Interactive survey	3.3

		syndromes and diseases, determine the need for additional examination, including specific genetic methods. Present the results of clinical , genetic and laboratory examination in the form of diaries and conclusions in the patient's medical history. Reasonably refer patients for medical and genetic counseling, provide the necessary documentation for this. Conduct preventive measures aimed at preventing hereditary and congenital diseases.			
4	Morphogenetic variants of development. Developmental defects	<p>Theoretical part: Morphogenetic variants of development (microanomalies, microsigns , stigmas of dysembryogenesis) and their significance in diagnostics of hereditary pathology. Congenital malformations (isolated, systemic and multiple). Etiological heterogeneity of congenital malformations. Genetic and environmental causes of teratogenesis. The concept of critical periods of ontogenesis. The concept of phenocopies.</p> <p>Practical part: Examine the patient for hereditary pathology. Assess the proband's phenotype based on the clinical and morphological examination data and make an assumption about the presence of congenital or hereditary pathology. Identify morphogenetic developmental variants during the examination.</p>	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	Current Testing Frontal survey Interactive survey	3.3
5	Chromosomal syndromes	<p>Theoretical part: Etiology and pathogenesis of chromosomal syndromes: numerical and structural changes. Factors causing chromosomal mutations: physical, chemical, biological; endogenous mutagens; mechanisms causing organohistogenesis disorders in chromosomal pathology. The place of chromosomal diseases in the structure of hereditary pathology, the number of known forms. General characteristics of chromosomal diseases. Features of the pathogenesis of chromosomal diseases. General issues of epidemiology and prevalence of chromosomal diseases. Cytogenetic methods for diagnosing chromosomal diseases and indications for cytogenetic examination of patients. Clinical genetics of chromosomal syndromes: Down, Patau , Edwards, Shereshevsky-Turner, Klinefelter , triplo -X, "chat cry", Wolf -Hirschhorn . Microdeletion syndromes (Prader-Willi, Angelman , Langer-Gideon).</p> <p>Practical part: Examine the patient for chromosomal</p>	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	Current Testing Frontal survey Interactive survey	3.3

		syndromes, recognize their common manifestations, diagnose congenital morphogenetic variants, and correctly use the appropriate terminology when describing the clinical picture (phenotype) of the patient. Collect anamnestic data and genealogical information, compile a pedigree, present it in graphic form, and analyze the inheritance of a disease or a disease trait in a family. Formulate a presumptive diagnosis of chromosomal pathology, determine the need for additional examination, including specific genetic methods. Present the results of clinical , genetic, and laboratory examination in the form of diaries and conclusions in the patient's medical history. Reasonably refer patients for medical and genetic counseling, provide the necessary documentation for conducting medical and genetic counseling.			
6	Monogenic hereditary diseases with autosomal dominant inheritance (Recklinghausen disease , Hamman - Rich disease, Minkowski-Chauffard anemia , Marfan syndrome , Ehlers-Danlos syndrome , achondroplasia , osteogenesis imperfecta , Alport syndrome)	<p>Theoretical part: Etiology, pathogenesis. General characteristics of monogenic pathology. Classification of Mendelian pathology. Clinical genetics of monogenic diseases with autosomal dominant inheritance (Recklinghausen disease, Hamman -Rich disease , Minkowski-Chauffard anemia , Marfan syndrome , Ehlers-Danlos syndrome , achondroplasia , osteogenesis imperfecta , Alport syndrome). Methods of diagnostics of monogenic diseases. General principles of treatment, rehabilitation and social adaptation of patients.</p> <p>Practical part: Examine the patient for a monogenic disease, recognize the manifestations of monogenic diseases, use terminology when describing the phenotype. Collect anamnestic data, compile a pedigree, present it in graphic form and analyze the hereditary disease (inheritance type). Formulate a presumptive diagnosis of a hereditary disease, determine the need for additional examination, including specific genetic methods (molecular genetic). Reasonably refer patients for medical and genetic counseling if a monogenic hereditary disease is suspected. Outline preventive measures aimed at preventing hereditary diseases of a monogenic nature.</p>	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	Current Testing Frontal survey Interactive survey	3.3
7	Monogenic hereditary diseases with	Theoretical part: Etiology, pathogenesis, general characteristics of monogenic pathology with autosomal recessive inheritance.	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6),	Current Testing	3.3

	<p>autosomal recessive inheritance (cystic fibrosis, celiac disease , adrenogenital syndrome, congenital hypothyroidism, Louis-Barr disease , Wilson-Konovalov disease, Kartagener disease)</p>	<p>Clinical genetics of monogenic diseases with autosomal recessive inheritance (cystic fibrosis, celiac disease , adrenogenital syndrome, congenital hypothyroidism, Louis - Barr syndrome , Wilson-Konovalov disease, Kartagener disease). Methods of diagnostics of monogenic diseases. General principles of treatment, rehabilitation and social adaptation of patients.</p> <p>Practical part: Examine the patient for a monogenic disease, recognize the manifestations of monogenic diseases, use terminology when describing the phenotype. Collect anamnestic data, compile a pedigree, present it in graphic form and analyze the hereditary disease (type of inheritance). Formulate a presumptive diagnosis of a hereditary disease, determine the need for additional examination, including specific genetic methods (molecular genetic). It is reasonable to refer patients for medical genetic counseling if a monogenic hereditary disease is suspected.</p> <p>To outline preventive measures aimed at preventing hereditary diseases of a monogenic nature.</p>	<p>OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .</p>	<p>Frontal survey Interactive survey</p>	
8	<p>Monogenic hereditary diseases with X-linked dominant inheritance (vitamin D-resistant rickets, Bloch-Sulzberger syndrome , follicular and pigmented keratosis, Conradi -Hunnerman syndrome , lipodystrophy with lipotrophic diabetes), X-linked recessive inheritance (hemophilia, muscular dystrophy) Duchenne -Becker, Lesch -Nyhan</p>	<p>Theoretical part: Etiology, pathogenesis, clinical genetics of monogenic diseases with X-linked dominant inheritance (vitamin D-resistant rickets, Bloch -Sulzberger syndrome , follicular and pigmented keratosis, Conradi-Hunnerman syndrome , partial lipodystrophy with lipotrophic diabetes). Etiology, pathogenesis, clinical genetics of monogenic diseases with X-linked recessive inheritance (hemophilia, myodystrophy) Duchenne -Becker, Lesch- Nyhan disease , color blindness, gout, testicular feminization syndrome). Etiology, pathogenesis, clinical genetics of monogenic diseases with Y -linked inheritance (male infertility). Etiology, pathogenesis, clinical genetics of monogenic diseases with mitochondrial inheritance (Leber optic atrophy , mitochondrial myopathy, Kearns-Sayre syndrome , benign tumors (oncocyomas), progressive ophthalmoplegia). Methods of diagnostics of monogenic diseases. General principles of treatment, rehabilitation and social adaptation of patients.</p>	<p>UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .</p>	<p>Current Testing Frontal survey Interactive survey</p>	3.3

	disease , color blindness, gout, testicular feminization syndrome), Y -linked inheritance (male infertility), mitochondrial inheritance (Leber's optic atrophy , mitochondrial myopathy, Kearns-Sayre syndrome , oncocytomas , ophthalmoplegia)	<p>Practical part: Examine the patient for a monogenic disease, recognize the manifestations of monogenic diseases, use terminology when describing the phenotype. Collect anamnestic data, compile a pedigree, present it in graphic form and analyze the hereditary disease (type of inheritance). Formulate a presumptive diagnosis of a hereditary disease, determine the need for additional examination, including specific genetic methods (molecular genetic). It is reasonable to refer patients for medical genetic counseling if a monogenic hereditary disease is suspected.</p> <p>To outline preventive measures aimed at preventing hereditary diseases of a monogenic nature.</p>			
9	Hereditary diseases of amino acid metabolism (phenylketonuria , tyrosinemia , alkaptonuria , leucinosi s , albinism, homocystinuria).	<p>Theoretical part: Etiology, pathogenesis, clinical genetics of hereditary amino acid metabolism diseases (phenylketonuria , tyrosinemia , alkaptonuria , leucinosi s , albinism, homocystinuria). Methods of diagnostics of hereditary amino acid metabolism diseases. Principles of treatment of hereditary amino acid metabolism diseases. Rehabilitation and social adaptation of patients with amino acid metabolism disorders.</p> <p>Practical part: Examine the patient for a monogenic disease, recognize the manifestations of monogenic diseases, use terminology when describing the phenotype. Collect anamnestic data, compile a pedigree, present it in graphic form and analyze the hereditary disease (type of inheritance). Formulate a presumptive diagnosis of a hereditary disease, determine the need for additional examination, including specific genetic methods (molecular genetic). It is reasonable to refer patients for medical genetic counseling if a monogenic hereditary disease is suspected. Outline preventive measures aimed at preventing hereditary diseases of a monogenic nature.</p>	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	Current Testing Frontal survey Interactive survey	3.3
10	Hereditary diseases of	<p>Theoretical part: Etiology, pathogenesis, clinical genetics of hereditary carbohydrate metabolism diseases (galactosemia , fructosemia , glycogenoses , mucopolysaccharidoses). Methods of diagnostics of hereditary carbohydrate metabolism diseases.</p>	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) ,	Current Testing Frontal survey Interactive	3.3

	carbohydrate metabolism (galactosemia , fructosemia , glycogenoses , mucopolysaccharidoses).	Principles of treatment of hereditary diseases of carbohydrate metabolism . Rehabilitation and social adaptation of patients with carbohydrate metabolism disorders. Practical part: Examine the patient for a monogenic disease, recognize the manifestations of monogenic diseases, use terminology when describing the phenotype. Collect anamnestic data, compile a pedigree, present it in graphic form and analyze the hereditary disease (type of inheritance). Formulate a presumptive diagnosis of a hereditary disease, determine the need for additional examination, including specific genetic methods (molecular genetic) . It is reasonable to refer patients for medical genetic counseling if a monogenic hereditary disease is suspected. Outline preventive measures aimed at preventing hereditary diseases of a monogenic nature.	PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	survey	
11	Hereditary diseases of lipid metabolism (Gaucher disease, Tay-Sachs , Niemann -Pick, Fabry disease, leukodystrophy , familial hypercholesterolemia).	Theoretical part: Etiology, pathogenesis, clinical genetics of hereditary lipid metabolism diseases (Gaucher disease, Tay-Sachs disease, Niemann -Pick disease, Fabry disease . leukodystrophy , familial hypercholesterolemia . Diagnostic methods and principles of treatment of hereditary lipid metabolism diseases . Rehabilitation and social adaptation of patients with lipid metabolism disorders. Practical part : Examine the patient for a monogenic disease, recognize the manifestations of monogenic diseases, use terminology when describing the phenotype. Collect anamnestic data, compile a pedigree, present it in graphic form and analyze the hereditary disease (type of inheritance). Formulate a presumptive diagnosis of a hereditary disease, determine the need for additional examination, including specific genetic methods (molecular genetic) . It is reasonable to refer patients for medical genetic counseling if a monogenic hereditary disease is suspected. Outline preventive measures aimed at preventing hereditary diseases of a monogenic nature.	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	Current Testing Frontal survey Interactive survey	3.3
12		Theoretical part: The proportion of multifactorially caused pathology in the structure of morbidity, disability and mortality of the population. The most common nosological forms (ischemic heart disease, hypertension, diabetes mellitus, peptic	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) ,	Current Testing Frontal survey Interactive	3.3

	Diseases with a hereditary predisposition (CHD, hypertension, diabetes, peptic ulcer, obesity, bronchial asthma) .	ulcer, obesity, bronchial asthma). Genetic and environmental factors of predisposition implementation. Risk factors and principles of identifying individuals with an increased risk of developing multifactorial diseases. Clinical and genealogical method in diagnosing multifactorial diseases and population screening. Ecogenetic diseases. Pharmacogenetics . Practical part: Examine a patient for multifactorial diseases, recognize the manifestations of this pathology, use terminology when describing the clinical picture. Collect anamnestic data and genealogical information, compile a pedigree, present it in graphic form and analyze the inheritance of a disease or a sign of a disease in a family. Formulate a presumptive diagnosis of multifactorial diseases, determine the need for additional examination, including specific genetic methods. Identify individuals with an increased risk of developing multifactorial diseases. Present the results of clinical , genetic and laboratory examination in the form of diaries and conclusions in the patient's medical history. Conduct preventive measures aimed at preventing multifactorial diseases and reducing their incidence.	PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	survey	
13	Prevention of hereditary diseases. Medical and genetic counseling.	Theoretical part: Types, ways and forms of prevention of hereditary diseases. Medical genetic counseling. Objectives, stages of implementation, content of stages of MGC. Prenatal diagnostics. Methods of prenatal diagnostics: invasive and non-invasive . Indications and contraindications for use, terms and conditions of implementation. Preconception prevention: essence, possible methods of implementation. Organization of medical genetic service in Russia. Legal and deontological issues in clinical genetics. Practical part: Prepare medical documentation and referral for medical and genetic counseling. Compile pedigrees of patients suffering from hereditary diseases with different types of inheritance. Determine risk factors and preventive measures for hereditary and congenital pathology.	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .		3.3
14	Treatment of	Theoretical part: Symptomatic treatment: medication, physiotherapy and surgery. Pathogenetic treatment (correction of metabolism at the substrate level, at the level of gene production,	OPK-1 (ID OPK-1.1. , 1.2), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) ,	Current Testing Frontal survey	3.3

	hereditary diseases. Gene therapy .	at the level of enzymes of hereditary and congenital diseases. Etiological treatment: principles and possibilities of gene therapy of hereditary and congenital diseases. Practical part: Justify the treatment tactics for a patient with a hereditary disease (at home, in a specialized children's institution, in a hospital).	PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	Interactive survey	
15	Credit	Checking the acquisition of competencies (interview on theoretical questions or situational tasks)	UK-1 (ID UK-1.1, 1.2, 1.5) ; OPK-1 (ID OPK-1.1. , 1.2), OPK-2, (ID OPK-2.2, 2.6), OPK-5 (ID OPK-5.2, 5.3, 5.4) PC-2 (PC ID 2.1, 2.2, 2.3, 2.4, 2.5) , PC-3 (PC ID 3.1, 3.2, 3.3, 3.4) , PC-5 (ID PC 5.2, 5.3) , PC-12 (ID PC 12.1, 12.2) .	Intermediate Testing Interview on theoretical issues	3.4

Explanation: Lesson No. 4 on the topic " Morphogenetic variants of development. Developmental defects" is held at the anatomical museum of the Department of Atomic and Operative Surgery of the Federal State Budgetary Educational Institution of Higher Education Amur State Medical Academy .

2. 4 Interactive forms of learning

In order to activate students' cognitive activity, interactive teaching methods are widely used in practical classes.

№ p /p	Topic of the practical lesson	Labor intensity in hours	Interactive form of learning	Labor intensity in hours, in % of the lesson
1	2	3	4	5
1	Introduction to Medical Genetics. Heredity and Pathology.	3.3	Interactive survey.	20 minutes (0.44 hours) / 13.1%
2	Semiotics of hereditary pathology, principles of clinical diagnostics. Clinical and morphological examination.	3.3	Interactive survey.	20 minutes (0.44 hours) / 13.1%
3	Methods of medical genetics. Screening programs. Neonatal screening.	3.3	Mutual reviews - taking notes.	20 minutes (0.44 hours) / 13.1%
4	Morphogenetic variants of development. Developmental defects.	3.3	Interactive survey.	20 minutes (0.44 hours) / 13.1%
5	Chromosomal syndromes.	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
6	Monogenic hereditary diseases with autosomal dominant inheritance (Recklinghausen disease , Hamman -Rich disease, Minkowski-Chauffard anemia , Marfan syndrome , Ehlers-Danlos syndrome , achondroplasia , osteogenesis imperfecta , Alport syndrome)	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
7	Monogenic hereditary diseases with autosomal recessive inheritance (cystic fibrosis, celiac disease , adrenogenital syndrome, congenital hypothyroidism, Louis- Barr disease , Wilson-Konovalov disease, Kartagener disease)	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
8	Monogenic hereditary diseases with X-linked dominant inheritance (vitamin D-resistant rickets, Bloch- Sulzberger syndrome , follicular and pigmented keratosis, Conradi-Hunerman syndrome , lipodystrophy with lipotrophic diabetes), X-linked recessive inheritance (hemophilia, muscular dystrophy) Duchenne -Becker, Lesch -Nyhan disease , color blindness, gout, testicular feminization syndrome), Y -linked inheritance (male infertility), mitochondrial inheritance (Leber optic atrophy , mitochondrial myopathy, Kearns-Sayre syndrome , oncocyomas , ophthalmoplegia).	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
9	Hereditary diseases of amino acid metabolism (phenylketonuria , tyrosinemia , alkaptonuria , leucinosia ,	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%

	albinism, homocystinuria).			
10	Hereditary diseases of carbohydrate metabolism (galactosemia , fructosemia , glycogenoses , mucopolysaccharidoses).	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
11	Hereditary diseases of lipid metabolism (Gaucher disease, Tay-Sachs , Niemann - Pick, Fabry disease, leukodystrophy , familial hypercholesterolemia).	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
12	Diseases with a hereditary predisposition (CHD, hypertension, diabetes, peptic ulcer, obesity, bronchial asthma) .	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
13	Prevention of hereditary diseases. Medical and genetic counseling.	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
14	Treatment of hereditary diseases. Gene therapy .	3.3	Analysis of clinical cases	20 minutes (0.44 hours) / 13.1%
15	Credit	3.4	Phenotype map defense, theoretical interview	20 minutes (0.44 hours) / 13.1%

2.5. Criteria for assessing students' knowledge

The basis for determining the level of knowledge, skills, and abilities are the assessment criteria - completeness and correctness:

- correct, precise answer;
- correct, but incomplete or inaccurate answer;
- incorrect answer;
- no answer.

When assigning marks, the classification of errors and their quality are taken into account:

- gross errors;
- similar errors;
- minor errors;
- shortcomings.

Distribution of marks in practical classes

No. p/p	Topic of the practical lesson	Theoretical part	Practical part	Overall rating	Forms of control
1	Introduction to Medical Genetics. Heredity and Pathology.	2-5	2-5	2-5	
2	Semiotics of hereditary pathology, principles of clinical diagnostics. Clinical and morphological examination.	2-5	2-5	2-5	Theoretical part Oral or written survey - Test tasks, including computer ones Practical part Interview on situational tasks, testing of practical skills at the patient's bedside, skills to work
3	Methods of medical genetics. Screening programs. Neonatal screening.	2-5	2-5	2-5	
4	Morphogenetic variants of development. Developmental defects.	2-5	2-5	2-5	
5	Chromosomal syndromes.	2-5	2-5	2-5	
6	Monogenic hereditary diseases with autosomal dominant inheritance (Recklinghausen disease , Hamman - Rich disease, Minkowski-Chauffard anemia , Marfan syndrome , Ehlers-Danlos syndrome , achondroplasia ,	2-5	2-5	2-5	

	osteogenesis imperfecta , Alport syndrome)				with regulatory documents - Performing exercises according to the model
7	Monogenic hereditary diseases with autosomal recessive inheritance (cystic fibrosis, celiac disease , adrenogenital syndrome, congenital hypothyroidism, Louis- Barr disease , Wilson-Konovalov disease, Kartagener disease)	2-5	2-5	2-5	
8	Monogenic hereditary diseases with X-linked dominant type of inheritance, X-linked recessive type of inheritance, Y -linked type of inheritance, mitochondrial type of inheritance.	2-5	2-5	2-5	
9	Hereditary diseases of amino acid metabolism (phenylketonuria , tyrosinemia , alkaptonuria , leucinosis , albinism, homocystinuria).	2-5	2-5	2-5	
10	Hereditary diseases of carbohydrate metabolism (galactosemia , fructosemia , glycogenoses , mucopolysaccharidoses).	2-5	2-5	2-5	
11	Hereditary diseases of lipid metabolism (Gaucher disease, Tay-Sachs , Niemann -Pick, Fabry disease, leukodystrophy , familial hypercholesterolemia).	2-5	2-5	2-5	
12	Diseases with a hereditary predisposition (coronary heart disease, hypertension, diabetes, peptic ulcer, obesity, bronchial asthma) .	2-5	2-5	2-5	
13	Prevention of hereditary diseases. Medical and genetic counseling.	2-5	2-5	2-5	
14	Treatment of hereditary diseases. Gene therapy .	2-5	2-5	2-5	
15	Credit	2-5	2-5	2-5	
Phenotype map		2-5			
Average score					

Rating scales for current/intermediate knowledge control

The success of students in mastering the discipline (topics/sections), practical skills and abilities is characterized by a qualitative assessment and is assessed on a 5-point system: “5” - excellent, “4” - good, “3” - satisfactory, “2” - unsatisfactory.

Test control evaluation criteria

"5" excellent - 90-100% correct answers

"4" is good - 80-89% correct answers

"3" satisfactory - 70-79% correct answers

"2" is unsatisfactory – less than 70% of correct answers.

Note : when passing the midterm test control on a discipline on the unified educational portal in the Moodle system , a student must score at least 80% correct answers to receive a positive grade.

Assessment criteria (grades) of the theoretical part

"5" - for the depth and completeness of mastery of the content of the educational material, in which the student easily navigates, for the ability to connect theoretical questions with practical ones, express and justify their judgments, correctly and logically present the answer; when testing, allows up to 10% of erroneous answers.

"4" - the student has fully mastered the educational material, is oriented in it, correctly states the answer, but the content and form have some inaccuracies; during testing, allows up to 20% of erroneous answers.

"3" - the student has mastered the knowledge and understanding of the main provisions of the educational material, but presents it incompletely, inconsistently, does not know how to express and justify his/her judgments; when tested, allows up to 30% of erroneous answers.

"2" - the student has fragmented and unsystematic knowledge of the educational material, is unable to distinguish between the main and the secondary, makes mistakes in defining concepts, distorts their meaning, presents the material in a disorderly and uncertain manner, and makes more than 30% of erroneous answers when tested.

Assessment criteria for the practical part

"5" - the student supervises a subject patient on a daily basis, has fully mastered the practical skills and abilities provided for by the course work program (correctly interprets the patient's complaints, anamnesis, objective examination data, formulates a clinical diagnosis, prescribes examination and treatment, interprets clinical, laboratory and instrumental indicators taking into account the norm).

"4" – the student supervises the subject patient on a daily basis, has fully mastered the practical skills and abilities provided for by the course work program, but allows for some inaccuracies.

"3" - the student does not regularly supervise the patient; the student has only some practical skills and abilities.

"2" - the student has visited the supervised patient less than 4 times, performs practical skills and abilities with gross errors.

Phenotype Map Evaluation Criteria

"5" – the design of the phenotype map meets the requirements;

"4" - in the phenotype map the student makes some inaccuracies in describing the examination data and in the wording of the conclusion.

"3" - the phenotype map is filled out with errors, written in illegible handwriting, and there are inaccuracies in the wording of the conclusion.

"2" - the phenotype map is written in illegible handwriting, and there are gross errors in the wording of the conclusion.

Working off disciplinary debts

If a student misses a class for a valid reason, he/she has the right to make it up and receive the maximum grade provided for by the course work program for that class. A valid reason must be documented.

If a student misses a class for an unjustified reason or receives a "2" mark for all activities in the class, he/she is required to make it up. In this case, the mark received for all activities is multiplied by 0.8.

If a student is excused from a class at the request of the dean's office (participation in sports, cultural and other events), then he is given a grade of "5" for this class, provided that he submits a report on the completion of mandatory extracurricular independent work on the topic of the missed class.

Assessment criteria for midterm assessment

Interim certification is carried out in 2 stages:

1. Test control in the " Moodle " system.
2. Answers to 2 questions.

Criteria for final assessment (midterm assessment)

Excellent - for the depth and completeness of mastering the content of the educational material, in which the student easily navigates, for the ability to connect theoretical questions with practical ones

, express and justify their judgments, correctly and logically present the answer; when testing, allows up to 10% of erroneous answers. Practical skills and abilities provided for by the working program of the discipline are fully mastered.

"Good" - the student has fully mastered the educational material, is oriented in it, correctly states the answer, but the content and form have some inaccuracies; during testing allows up to 20% of erroneous answers. Completely practical skills and abilities provided by the working program of the discipline, but allows some inaccuracies

"Satisfactory" - the student has mastered the knowledge and understanding of the main provisions of the educational material, but presents it incompletely, inconsistently, does not know how to express and justify his/her judgments; during testing, allows up to 30% of erroneous answers. Has only some practical skills and abilities.

"Unsatisfactory" - the student has fragmented and unsystematic knowledge of the educational material, is unable to distinguish between the main and secondary, makes mistakes in defining concepts, distorts their meaning, presents the material in a disorderly and uncertain manner, and makes more than 30% of erroneous answers during testing. Performs practical skills and abilities with gross errors.

Academic rating of students

The rating indicator for a discipline is formed on the basis of the assessment of the student's knowledge, skills, abilities based on the results of the midterm assessment and bonus/penalty points. The maximum result that a student can achieve is 10 points (5 points for the midterm assessment + 5 bonus points), the minimum is 0 points.

Distribution of bonus points (no more than 5 points per discipline):

- **1 point** – oral presentation at conferences;
- **0.25 points** – poster presentation at conferences;
- **1 point** – winner of the Olympiad (prize places);
- **0.25 points** – participant in the Olympiad;
- **1 point** – active participation of the student in career guidance and educational work
- **0.5 points** – student participation in the work of the student scientific society on pediatrics
- **0.5 points** – additional performance of independent extracurricular work (presentations, literature review, duty, etc.)
- **0.5 points** – the student's diligent attitude to learning (no absences from lectures or classes for no good reason; active participation of the student in all classes).

Distribution of penalty points (no more than 5 points per discipline):

- absences from lectures and practical classes for an unjustified reason – **1 point**;
- damage to cathedral property – **1 point**;
- disrespectful attitude towards the teacher, patients, medical staff . personnel – **1 point**;
- unkempt appearance, lack of a robe **-0.5 points**;
- systematic unpreparedness for classes, failure to complete independent work within the specified time frame – **0.5 points**;
- violation of class discipline – **1 point**.

If a student has a grade of "5", "4", "3" for the midterm assessment, then he receives a "pass" for the discipline Medical Genetics; if a student has a grade of "2" for the midterm assessment, then he receives a "fail" for the discipline Medical Genetics.

2.6. Independent work of students: in-class and out-of-class

The main didactic tasks of independent work of students under the guidance of a teacher: consolidation of knowledge and skills acquired in the process of studying the academic discipline in lectures and practical classes; prevention of their forgetting; expansion and deepening of educational material; formation of the ability and skills of independent work; development of independence of thinking and creative abilities of students.

Independent classroom work of students

Independent classroom work of students makes up 25% of the time allocated for the lesson. The main didactic tasks of independent work of students under the guidance of a teacher: consolidation of knowledge and skills acquired in the process of studying the academic discipline in lectures and practical classes; prevention of their forgetting; expansion and deepening of the educational material; formation of the ability and skills of independent work; development of independence of thinking and creative abilities of students.

Students' independent classroom work includes:

1. Supervision of thematic patients.
2. Preparation of a plan for examination, including specific molecular genetic, cytogenetic studies, and treatment.
3. Compilation of pedigrees of patients suffering from hereditary diseases with different types of inheritance.
4. Determination of risk factors and measures for the prevention of hereditary and congenital pathologies.
5. Calculation of the prognosis for the occurrence of hereditary pathology in the family of the person examined.
6. Determination of measures to prevent hereditary and congenital pathology in the proband's family.
7. Compilation of a differential diagnostic table of the main chromosomal diseases indicating the karyotype.
8. Work with medical documentation (orders, genetic map).
9. Solving situational problems.
10. Working with the genetic dictionary.
11. Compilation of a pedigree for a patient with a multifactorial disease with calculation of the Makarov index.
12. Determining the prognosis of hereditary pathology for a specific patient.

Extracurricular independent work of students

For independent extracurricular work the following may be used: studying the main and additional educational and scientific literature; solving situational problems, test assignments, working in an online classroom, preparing oral reports; writing essays. This type of educational activity should be based on the students' activity, initiative, consciousness and self-activity.

Organization of extracurricular independent work of students.

	Topic	Time to prepare for the lesson	Forms of extracurricular independent work of students	
			Mandatory and the same for all students	At the student's choice
1.	Introduction to Medical Genetics. Heredity and Pathology.	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom .	Report on the topic: "Contribution domestic scientists in the development of medical genetics"
2.	Semiotics of hereditary pathology, principles of clinical diagnostics.	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations,	Creation of a computer presentation on the topic "Portrait diagnostics of chromosomal syndromes", "Minor developmental

	Clinical and morphological examination.		abstracting, writing notes), workbook preparation, work in an online classroom .	anomalies"
3.	Methods of medical genetics. Screening programs. Neonatal screening.	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom .	Creation of a computer presentation on the topic "Methods of molecular genetic diagnostics", "Cytogenetic method"
4.	Morphogenetic variants of development. Developmental defects.	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom .	Abstract on the topic "Teratogenic factors"
5.	Chromosomal syndromes.	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom .	Drawing up a situational task on Down syndrome, Shereshevsky-Turner syndrome, Klinefelter syndrome . Abstract on the topic "Neoplasia in chromosomal diseases" Creation of a computer presentation on the topic "Chromosomal syndromes", "Diagnostics of chromosomal syndromes"
6.	Monogenic hereditary diseases with autosomal dominant inheritance (Recklinghausen disease , Hamman - Rich disease, Minkowski-Chauffard anemia , Marfan syndrome , Ehlers-Danlos syndrome , achondroplasia , osteogenesis imperfecta , Alport syndrome)	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom .	Drawing up a situational task on Marfan's disease . Review of periodicals, internet sources on the topic "Hereditary diseases of the respiratory system", "Hereditary diseases of the nervous system" Creating a computer presentation on the topic " Neurofibromatosis ", " Imperfect osteogenesis »
7.	Monogenic hereditary diseases with autosomal	2.6	Preparation on theoretical issues (lecture reading, primary and secondary	Drawing up a situational task on cystic fibrosis , celiac disease .

	recessive inheritance (cystic fibrosis, celiac disease, adrenogenital syndrome, congenital hypothyroidism, Louis- Barr disease, Wilson-Konovalov disease, Kartagener disease)		literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom .	Review of periodicals, internet sources on the topic "Adrenogenital syndrome", "Congenital hypothyroidism". Creation of a computer presentation on the topic " Louis- Barr syndrome , Wilson-Konovalov disease, Kartagener disease ".
8	Monogenic hereditary diseases with X-linked dominant inheritance (vitamin D-resistant rickets, Bloch- Sulzberger syndrome , etc.), X-linked recessive inheritance (hemophilia, muscular dystrophy Duchenne -Becker, Lesch -Nyhan disease , color blindness, gout), Y-linked type of inheritance (male infertility), mitochondrial type of inheritance (Leber optic atrophy , mitochondrial myopathy, Kearns-Sayre syndrome).	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom	Drawing up a situational task on vitamin D-resistant rickets, Bloch- Sulzberger syndrome . Review of periodicals, internet sources on the topic " Conradi-Hunerman syndrome ", " Lipodystrophy with lipotrophic diabetes". Creation of a computer presentation on the topic "Hemophilia, myodystrophy Duchenne -Becker, Lesch - Nyhan disease, Leber's optic atrophy , mitochondrial myopathy, Kearns-Sayre syndrome .
9	Hereditary diseases of amino acid metabolism (phenylketonuria , tyrosinemia , alkaptonuria , leucinosis , albinism, homocystinuria).	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom	Drawing up a situational task on amino acid metabolism diseases. Review of periodicals, internet sources on the topic "Hereditary disorders of amino acid metabolism", Creation of a computer presentation on the topic "Hereditary disorders of amino acid metabolism".
10	Hereditary diseases of carbohydrate	2.6	Preparation on theoretical issues (lecture reading,	Drawing up a situational task on galactosemia .

	metabolism (galactosemia , fructosemia , glycogenoses , mucopolysaccharidoses)		primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom	Review of periodicals, internet sources on the topic "Hereditary disorders of carbohydrate metabolism" Creation of a computer presentation on the topic " Glycogenoses "
11	Hereditary diseases of lipid metabolism (Gaucher disease, Tay-Sachs , Niemann -Pick, Fabry disease, leukodystrophy , familial hypercholesterolemia).	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom	Compilation of a situational task on Gaucher disease. Review of periodicals, Internet sources on the topic "Hereditary disorders of lipid metabolism". Creation of a computer presentation on the topic " Lipidoses ".
12.	Diseases with a hereditary predisposition (coronary heart disease, hypertension, diabetes, peptic ulcer, obesity, bronchial asthma) .	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom .	Review of periodicals, internet sources on the topic "Genetic aspects of carcinogenesis", "Genetic predisposition to bronchial asthma".
13.	Prevention of hereditary diseases. Medical and genetic counseling.	2.6	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation , work in an online classroom .	Creation of a computer presentation on the topic "Methods of prenatal diagnostics". Review of periodicals, internet sources on the topic "Medical and genetic counseling"
14.	Treatment of hereditary diseases. Gene therapy .	2,2	Preparation on theoretical issues (lecture reading, primary and secondary literature, methodological recommendations, abstracting, writing notes), workbook preparation, work in an online classroom .	Creation of a computer presentation on the topic " Gene therapy ", Report on the topic "Replacement therapy"
Labor intensity in hours		36	24	12
Total labor intensity in hours		36		

2.7. Research (project) work

Research (project) work of students (RWS) is a mandatory section of the study of the discipline and is aimed at the comprehensive formation of general cultural and professional competencies of

students and involves the study of specialized literature and other scientific and technical information on the achievements of domestic and foreign science and technology in the relevant field of knowledge, participation in scientific research, etc.

The topic of research work can be chosen by students independently in consultation with the teacher or from the list below (taking into account the scientific direction of the department).

Sample research topics:

3. Malabsorption syndrome.
4. Cystic fibrosis.
5. Celiac disease.
6. Connective tissue dysplasia syndrome.
7. Orphan pathology.
8. Adrenogenital syndrome.
9. Chromosomal pathology.

The student can present the results of these studies at meetings and the final conference of the Student Scientific Society, at conferences; and publish them in a scientific and practical publication.

III. EDUCATIONAL, METHODOLOGICAL, LOGISTICAL AND INFORMATIONAL SUPPORT FOR THE DISCIPLINE:

3.1 Main literature:
1. Azova, M. M. General and medical genetics. Tasks : a tutorial / edited by M. M. Azova . - Moscow : GEOTAR-Media, 2021. - 160 p. - 160 p. - ISBN 978-5-9704-5979-9. - Text : electronic (date accessed: 05/04/2021). - Access mode : by subscription. http://www.studmedlib.ru/book/ISBN9785970459799.html
2. Bochkov, N. P. Clinical genetics : textbook / edited by Bochkov N. P. - Moscow: GEOTAR-Media, 2020. - 592 p. - ISBN 978-5-9704-5860-0. - Text : electronic (date accessed: 05/04/2021). - Access mode : by subscription. http://www.studmedlib.ru/book/ISBN9785970458600.html
3.2 Further reading:
1. Akulenko, L. V. Medical genetics: textbook . p individual / Akulenko L. V. et al. - Moscow: GEOTAR-Media, 2015. - 192 p. - ISBN 978-5-9704-3361-4. - Text : electronic // URL: http://www.studmedlib.ru/book/ISBN9785970433614.html (date accessed: 06.05.2021). - Access mode : by subscription.
2. Babtseva A.F. Medical genetics: textbook / A.F. Babtseva, O.S. Yutkina, E.B. Romantsova . - Blagoveshchensk, 2012.-166 p.-Direct text.

3.3. Educational and methodological care disciplines prepared by the staff of the department

Textbooks with the stamp (Coordinating Council for the field of education "Healthcare and Medical Sciences"):

Babtseva A.F., Yutkina O.S., Romantsova E.B. Textbook with the stamp of the UMO "Medical Genetics", Blagoveshchensk, 2011.

Electronic and digital technologies:

1. Online course on the subject "Medical Genetics" in the EIS FGBOU VO Amur State Medical Academy

Access mode: <https://educ-amursma.ru/course/view.php?id=546>

Multimedia materials on electronic media (CD , DVD)

Electronic version of the video:

"Morphogenetic variants of development. Congenital malformations."

Electronic version of the educational film:

"Medical Genetics. Overview of the Curriculum"

"Gene diseases"

"Chromosomal syndromes"

"Methods of studying hereditary diseases"

"Multifactorial diseases. Prevention of hereditary pathology. Fundamentals of medical and genetic counseling."

**Video films, photographs used in teaching students:
at the department (CDs)**

1. "Hereditary metabolic diseases";
2. "Treatment of hereditary metabolic diseases";
3. "Semiotics of hereditary diseases"
4. "Neonatal screening";
5. "Molecular genetic diagnostic method";
6. " Phenylketonuria ";
7. "Chromosomal diseases";
8. "Clinical-genealogical method";
9. "Population method";
10. " Galactosemia ";
11. "Congenital adrenal hyperplasia";
12. "Adrenogenital syndrome";
13. "Cystic fibrosis";
14. "Cytogenetic method";
15. " Prenatal diagnosis";
16. "Genetics of congenital malformations";
17. " Gene therapy ";
18. "Medical and genetic counseling";
19. "Congenital hypothyroidism"
20. "Congenital defects of the urinary system in children"
21. "Multifactorial diseases"
22. "Monogenic diseases"
23. "Celiac disease"
24. "Principles of treatment and prevention of hereditary diseases "

List of albums, stands, tables, tablets, handouts used in training (prepared by the department staff)

Photo albums (developmental defects) – 1;

Slides for the overhead projector : "Chromosomal diseases. Portrait diagnostics". **Stands** :

"Introduction to medical genetics", "Methods of medical genetics", "Hereditary diseases", " Prenatal diagnostics".

Posters : "Classification of hereditary diseases", "Basics of symmetry".

Folders-booklets:

1. "Introduction to Medical Genetics. Heredity and Pathology";
2. "Semiotics of hereditary pathology and principles of clinical diagnostics. Family as an object of medical and genetic observation";
3. "Morphogenetic variants of development. Developmental defects";
4. "Methods of medical genetics";
5. "Chromosomal diseases. Chromosomal imprinting. Family predisposition";
6. "Monogenic hereditary diseases";
7. Maldigestion and malabsorption syndrome ";
8. "Multifactorial";

9. "Prevention of hereditary diseases. Medical and genetic counseling. Prenatal diagnostics. Screening programs. Environmental protection";
10. "Treatment of hereditary diseases. Gene therapy ."

Chromosomal diseases

1. Trisomy of the long arm of chromosome 14 (14q+ syndrome) ;
2. Orbeli syndrome;
3. Cri du chat syndrome;
4. Down syndrome;
5. Chromosome 18- q syndrome (short arm deletion);
6. Turner's disease ;
7. Hirschhorn syndrome (2 pcs)
8. Trisomy 9 syndrome ;
9. Deletion of chromosome 10p (2 pcs)
10. Trisomy 10 syndrome ;
11. Syndrome 11 q trisomy (2 pcs);
12. Jacobsen syndrome (chromosome 11 q - syndrome);
13. 14q + syndrome ;
14. Klinefelter syndrome ; Patau syndrome ; Edwards syndrome;

Genetic pathology

1. Celiac disease (2 pcs)
2. Phenylketonuria (2 pieces)
3. Gilbert's syndrome
4. Wolman's disease (2 pcs)
5. Hyperkalemic form of parkinsonism myoplegia (Gamsthorp's disease)
6. Hamman -Rich disease
7. Crigler-Najjar syndrome
8. Myasthenia
9. Neurofibromatosis
10. Goodpasture's syndrome
11. Louis-Bar syndrome
12. Huntington's chorea
13. Anaphalipoproteinemia (Tangier's disease)
14. Menkes syndrome
15. Myodystrophy Duchenne -Becker
16. Myotonic dystrophy (Steinert's disease , dystrophic myotonia)
17. Parkinson's disease
18. Kartagener's syndrome (2 pieces)
19. Hereditary Pierre-Marie's ataxia
20. Congenital myotonia (Leiden- Thomsen disease)
21. Wilson-Konovalov disease (2 pcs)
22. Nyhan syndrome
23. Lowe's syndrome
24. Thalassemia
25. Cystinosis
26. Stickler syndrome
27. Osler - Rendu -Weber disease
28. Vitamin D resistant rickets
29. Marfan syndrome
30. Cystic fibrosis
31. Sickle cell anemia
32. Galactosemia (3 pcs)
33. Hemophilia

34. Ehlers-Danlos syndrome
35. Osteogenesis imperfecta (disease) Lobshtein-Vrolika)
36. Hereditary spherocytosis
37. De Toni- Debreu - Fanconi syndrome
38. Fibrodysplasia ossifying progressive
39. Achondroplasia
40. Crohn's disease
41. Alkaptonuria
42. Mucopolysaccharidosis , type 6 (Maroteau -Lami syndrome) (3 pieces)
43. Metachromatic leukodystrophy
44. Leigh syndrome
45. Kearns-Sayre syndrome
46. Mucopolysaccharidosis type 2 (Hunter syndrome)
47. Familial hypercholesterolemia
48. Mucopolysaccharidosis type 8 (Di syndrome Ferrante)
49. Fabry disease
50. Niemann -Pick disease
51. Mucopolysaccharidosis type 3 (Sanfilippo syndrome)

Handouts: case histories, situational tasks, test assignments, albums on the topics being studied, standards for providing specialized assistance and protocols on the topics being discussed.

3.4. Equipment used for the educational process

No.	Name	Quantity
1	Head of Department's Office	1
	Table	2
	Chairs	16
	Bookshelf	5
2	Assistant	1
	Table	3
	Chairs	4
	Bookshelf	2
	Wardrobe	1
3	Study room #1-4	4
	Table	21
	Chairs	66
	Stands	9
	Board	2
	Bookshelf	4
	Visual aids	42
4	Personal computer	1
5	Laptop	1
6	Multimedia video projector	2
7	Screen	1
8	printer/copier	1
9	Equipment of the laboratory of the State Autonomous Healthcare Institution of the Arkhangelsk Region City Clinical Hospital (clinical, immunological) Equipment of the functional diagnostics department of the State Autonomous Healthcare Institution of the Arkhangelsk Region City Clinical Hospital	

	<p>Equipment of the X-ray room of the State Autonomous Healthcare Institution of the Arkhangelsk Region, the Children's City Clinical Hospital</p> <p>Departments of the State Autonomous Healthcare Institution of the Arkhangelsk Region Children's City Clinical Hospital: pediatrics, day hospital, neurology, orthopedics, restorative and rehabilitation treatment</p>	
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3.5. Professional databases, information and reference systems, electronic educational resources

Item No	Resource name	Resource Description	Access	Resource address
Electronic library systems				
1	"Student consultant. Electronic library of the medical university"	For students and teachers of medical and pharmaceutical universities. Provides access to electronic versions of textbooks, teaching aids and periodicals .	library, individual access	http://www.studmedlib.ru/
2	PubMed	Free search engine in the largest medical bibliographic database MedLine . Documents medical and biological articles from the specialized literature, and also provides links to full-text articles.	library, free access	http://www.ncbi.nlm.nih.gov/pubmed/
3	Oxford Medicine Online	A collection of Oxford medical publications, bringing together over 350 titles into a single, cross-searchable resource. Publications	library, free access	http://www.oxfordmedicine.com

		include The Oxford Handbook of Clinical Medicine and The Oxford Textbook of Medicine , electronic versions of which are constantly updated.		
4.	Human Biology Knowledge Base	Reference information on physiology , cell biology , genetics , biochemistry , immunology , pathology . (Resource of the Institute of Molecular Genetics of the Russian Academy of Sciences .)	library, free access	http://humbio.ru/
5 .	Medical online library	Free reference books, encyclopedias, books, monographs, abstracts, English-language literature, tests.	library, free access	http://med-lib.ru/
Information systems				
6.	Russian Medical Association	Professional Internet resource . Objective: to promote effective professional activity of medical personnel. Contains the charter, personnel, structure, rules of entry, information about the Russian Medical Union	library, free access	http://www.rmass.ru/
7.	Web -medicine	The site presents a catalog of professional medical resources, including links to the most authoritative subject sites, journals, societies,	library, free access	http://webmed.irkutsk.ru/

		as well as useful documents and programs. The site is intended for doctors, students, employees of medical universities and scientific institutions.		
Databases				
8.	World Health Organization	The site contains news, statistics on countries that are members of the World Health Organization, fact sheets, reports, WHO publications and much more.	library, free access	http://www.who.int/ru/
9.	Ministries of Science and higher education Russian Federation	Website of the Ministry of Science and Higher Education Russian Federation contains news, newsletters, reports, publications and much more	library, free access	http://www.minobrnauki.gov.ru
10.	Ministry of Education Russian Federation	Website of the Ministry of Education of the Russian Federation contains news, newsletters, reports, publications and much more	library, free access	https://edu.gov.ru/
11.	Federal portal "Russian education"	Single window of access to educational resources. This portal provides access to textbooks on all branches of medicine and health care	library, free access	http://www.edu.ru/ http://window.edu.ru/catalog/?p_rubr=2.2.81.1
Bibliographic bases data				
12.	Database "Russian Medicine"	It is created in the Central Scientific and Methodological	library, free access	http://www.scsml.rssi.ru/

		Library and covers the entire collection, starting from 1988. The database contains bibliographic descriptions of articles from domestic journals and collections, dissertations and their abstracts, as well as domestic and foreign books, collections of institute proceedings, conference materials, etc. Thematically, the database covers all areas of medicine and related areas of biology, biophysics, biochemistry, psychology, etc.		
13	eLIBRARY.RU	Russian information portal in the field of science, technology, medicine and education, containing abstracts and full texts of more than 13 million scientific articles and publications. Powered by eLIBRARY . RU Electronic versions of more than 2,000 Russian scientific and technical journals are available, including more than 1,000 open access journals	library, free access	http://elibrary.ru/defaultx.asp
14	Portal Electronic library of dissertations	Currently, the Electronic Library of Dissertations of the Russian State	library, free access	http://diss.rsl.ru/?menu=disscatalog/

		Library contains more than 919,000 full texts of dissertations and abstracts.		
15.	Medline .r u	Medical and biological portal for specialists. Biomedical journal. Last updated February 7, 2021.	library, free access	http://www.medline.ru

3.6. Licensed and freely distributed software used in the educational process

I. Commercial software products		
1.	Operating system MS Windows 7 Pro	License number 48381779
2.	Operating system MS Windows 10 Pro , MS Office	AGREEMENT No. 142 A dated December 25, 2019
3.	MS Office	License number: 43234783, 67810502, 67580703, 64399692, 62795141, 61350919
4.	Kaspersky Endpoint Security for Business Advanced	Agreement No. 977/20 dated 12/24/2020
5.	IC : University PROF	LICENSE AGREEMENT No. 2191 dated 15.10.2020
6.	PROF Library	LICENSE AGREEMENT No. 2281 dated 11.11.2020
II. Freely distributed software		
1.	Google Chrome	Freely distributed Distribution conditions: https://play.google.com/about/play-terms/index.html
2.	Browser « Yandex »	Freely distributed License agreement for the use of the Yandex Browser software https://yandex.ru/legal/browser_agreement/
3.	Dr.Web CureIt !	Freely distributed License Agreement: https://st.drweb.com/static/new-www/files/license_CureIt_ru.pdf
4.	OpenOffice	Freely distributed License: http://www.gnu.org/copyleft/lesser.html
5.	LibreOffice	Freely distributed License: https://ru.libreoffice.org/about-us/license/

3.7. Resources of the information and telecommunications network "Internet"

Standards for the provision of primary health care

<https://www.rosminzdrav.ru/ministry/61/22/page-979/page-983/1-standard-primary-medical-sanitary-assistance>

Standards for the provision of specialized medical care

<https://www.rosminzdrav.ru/ministry/61/22/page-979/page-983/2-standard-specialized-medical-assistance>

Procedures for providing medical care to the population of the Russian Federation

<https://www.rosminzdrav.ru/ministry/61/4/page-857/services-services-medical-assistance-national-federation>

Ministry of Health of the Amur Region (documents)

<https://amurzdrav.ru/%D0%B4%D0%BE%D0%BA%D1%83%D0%BC%D0%B5%D0%BD%D1%82%D1%8B/>

Federal Electronic Medical Library

<http://www.femb.ru>

Union of Pediatricians of Russia (Clinical Guidelines)

<http://www.pediatr-russia.ru> - <http://www.pediatr-russia.ru/news/recommend>

Pediatrics Magazine

<http://pediatrajournal.ru>

Magazine « Consilium medicus »

<http://con-med.ru/magazines/pediatrics>

Journal "Russian Bulletin of Perinatology and Pediatrics"

<http://www.ped-perinatology.ru>

Magazine "Attending physician"

<http://www.lvrach.ru>

"Russian Medical Journal"

<http://www.rmj.ru>

IV . ASSESSMENT TOOLS FUND

4.1 Current test control (input, initial), final

4.1.1 Examples of test tasks for entrance knowledge assessment (with standard answers)

Conducted on a single information and educational portal in the Moodle system <https://educ-amursma.ru/mod/quiz/view.php?id=18393> total number of test tasks – 50.

1. MITOSIS IS OF GREAT BIOLOGICAL IMPORTANCE BECAUSE

1. It underlies the mechanism of gamete formation
2. Is a source of combinatorial variability
3. Provides a change in the original chromosome set
4. Ensures uniform distribution of hereditary material between daughter cells
5. Ensures the replication of the hereditary material of the mother cell

2. TRANSLATION OF THE TERM MEANING "PARALYSIS OF ONE LIMB":

1. monoplegia
2. panplegia
3. diplegia
4. hemiplegia

STANDARD ANSWERS TO TEST TASKS 1-4. 2-1.

4.1.2 Examples of test tasks for initial knowledge control (with sample answers)

Access mode <https://educ-amursma.ru/course/view.php?id=213>

Please indicate one correct answer.

1 . SUBJECT OF STUDY OF MEDICAL GENETICS

- 1) animal diseases
- 2) hereditary diseases of humans

- 3) human infectious diseases
- 4) diseases of the neonatal period
- 5) injuries

2. HEREDITARY PATHOLOGY IS CHARACTERIZED BY

- 1) multiple organ involvement, resistance to therapy
- 2) acute course
- 3) favorable outcome of the disease
- 4) lack of chronization of the process

STANDARD ANSWERS TO TEST TASKS 1-2. 2-1.

4.1.3. Examples of test tasks for the final knowledge assessment (with sample answers)

Conducted by the Moodle system (e-mail address <https://educ-amursma.ru/mod/quiz/view.php?id=18394>).

Total number of test tasks – 100.

1. A DIPLOID SET OF CHROMOSOMES IS FORMED AS A RESULT OF

- 1) meiosis
- 2) mitosis
- 3) division of autosomes
- 4) division of sex chromosomes

2. A HAPLOID SET OF CHROMOSOMES IS FORMED AS A RESULT

- 1) meiosis
- 2) mitosis
- 3) division of autosomes
- 4) division of sex chromosomes

SAMPLE ANSWERS TO TEST QUESTIONS 1-3 2-4

4.2 SITUATIONAL PROBLEMS (WITH STANDARD ANSWERS)

Task 1.

The mother of a 6.5-month-old girl consulted a pediatrician complaining that her child was not rolling over, sitting, or standing. Past medical history: 1 pregnancy without any complications, 1 term birth, birth weight 2900. Breastfeeding. By 1.5 months, she began to hold her head up, and by 2.5–3 months, she has been rolling over from her back to her stomach and back. Past medical history: The child began vomiting at 3.5 months, began to refuse food, and by the 4th month, her weight had increased by 100 mg (with the norm being 700 mg). At 4 months, she had an acute respiratory viral infection complicated by obstructive syndrome and was treated in hospital. After being discharged from the hospital, the mother noticed that the girl had become less active, had stopped rolling over, showing interest in toys, and recognizing loved ones. Objective examination: the child's condition is serious. Skin is coffee-yellow. Signs of hypotrophy (weight deficit of 18%). In the lungs, puerile breathing is conducted in all fields. Heart sounds are rhythmic. The abdomen is soft, the edge of the liver is palpated (protrudes from under the edge of the costal arch by 3 cm) of a dense - elastic consistency. The spleen protrudes from under the edge of the costal arch by 1,5 cm. Muscle hypotonia. Does not sit independently, does not follow a toy, gaze is fixed.

1. What illness should we think about in a child?
2. What additional tests are needed to confirm the diagnosis?
3. What is the prognosis for this child?
4. Prognosis for subsequent offspring of this married couple.

Standard solution to problem #1

1. One should think about a hereditary disease associated with lipid metabolism disorders (storage diseases) – Tay-Sachs disease.
2. Consultation with a neurologist; examination of the fundus (cherry pit symptom); examination of the organs of vision and hearing; collection of information and preparation of a pedigree; identification of heterozygous carriers in parents; DNA diagnostics.
3. The prognosis is unfavorable, with death occurring by age 3.

4. The inheritance pattern of the disease is autosomal recessive: a sick child is born into a family where both parents are carriers of the mutant gene. Prognosis for subsequent children: 25% - probability of having a healthy child; 25% - a sick child; 50% - a phenotypically healthy carrier of the mutant gene.

4.3 List of practical skills that a student should have after mastering the discipline

1. Obtain information about sick and healthy family members based on a survey, examination and available medical documents.
2. Conduct an objective examination of the proband's organs and systems, and an examination of the parents and other relatives.
3. Diagnose syndromes requiring emergency care, determine its volume and sequence.
4. Refer for special studies (biochemical, cytogenetic, molecular genetic, ultrasound, etc.).
5. Establish a preliminary diagnosis and conduct differential diagnostics of hereditary diseases.
6. Identify individuals with increased risk of developing multifactorial diseases.
7. Determine indications for referral for consultation to doctors of other specialties.
8. Justify the treatment tactics for a patient with a hereditary disease (at home, in a specialized children's institution, in a hospital).
9. Evaluate the results of laboratory and special diagnostic methods for hereditary diseases.
10. Prepare medical documentation and referrals for medical and genetic counseling.
11. Examine the patient for hereditary pathology, recognize common manifestations of hereditary pathology, diagnose congenital morphogenetic variants, correctly use the appropriate terminology when describing the clinical picture (phenotype) of the patient.
12. Conduct preventive measures aimed at preventing hereditary and congenital diseases, reducing the incidence of widespread diseases of multifactorial nature.

4. 4 List of questions for the test

1. Medical genetics. Subject and tasks of medical genetics.
2. History of the development of medical genetics.
3. Etiology of hereditary diseases. Mutations.
4. Classification of hereditary diseases. Pathogenesis of hereditary diseases (molecular, cellular, tissue levels).
5. Semiotics of hereditary pathology, principles of clinical diagnostics.
6. Clinical and morphological examination. Stigmas of dysembryogenesis .
7. Stages of diagnosis of hereditary diseases.
8. Signs of hereditary diseases.
9. Screening programs. Neonatal screening.
10. Clinical-genealogical method. Definition, essence of the method, indications for its use.
11. Cytogenetic method. Definition, essence of the method, indications for its use.
12. Biochemical method. Definition, essence of the method, indications for its use.
13. Molecular genetic method. Definition, essence of the method, indications for its use.
14. Classification of teratogens . Critical periods of the intrauterine period of ontogenesis.
15. Developmental defects.
16. Classification and general characteristics of chromosomal diseases.
17. Autosome anomalies. Down, Patau , Edwards syndrome.
18. Sex chromosome abnormalities. Turner-Sherevsky syndrome, Klinefelter syndrome .
19. Recklinghausen's disease .
20. Minkowski-Choffar anemia .
21. Marfan syndrome .
22. Achondroplasia .
23. Osteogenesis imperfecta .
24. Alport syndrome .
25. Cystic fibrosis.

26. Celiac disease.
27. Adrenogenital syndrome.
28. Congenital hypothyroidism.
29. Barr syndrome .
30. Wilson-Konovalov disease.
31. Kartagener's disease .
32. Monogenic hereditary diseases with an X-linked dominant type of inheritance (vitamin D-resistant rickets, Bloch- Sulzberger syndrome).
33. Monogenic hereditary diseases with X-linked recessive inheritance (hemophilia, myodystrophy Duchenne -Becker).
34. Monogenic hereditary diseases with Y -linked inheritance (male infertility),
35. Monogenic hereditary diseases with mitochondrial inheritance (Leber's optic atrophy , mitochondrial myopathy).
36. Hereditary diseases of amino acid metabolism (phenylketonuria , homocystinuria).
37. Hereditary diseases of carbohydrate metabolism (galactosemia , fructosemia).
38. Hereditary diseases of carbohydrate metabolism (glycogenoses).
39. Hereditary diseases of carbohydrate metabolism (mucopolysaccharidoses).
40. Hereditary diseases of lipid metabolism (Gaucher disease, Tay-Sachs disease).
41. Hereditary diseases of lipid metabolism (Niemann -Pick disease, Fabry disease).
42. Hereditary diseases of lipid metabolism (leukodystrophy , familial hypercholesterolemia).
43. Genetic and environmental risk factors for the development of diseases with a hereditary predisposition.
44. Prevention of hereditary diseases. Preconception preparation.
45. Medical genetic counseling: objectives and stages of implementation.
46. Prenatal diagnostics. Methods of prenatal diagnostics.
47. Pathogenetic treatment of hereditary diseases.
48. Symptomatic treatment of hereditary diseases.
49. Etiological treatment of hereditary diseases.
50. Deontological and ethical issues of medical genetics.

APPROVED
at a department meeting
childhood diseases

Protocol No. 9 of 22. 04.2026
Head of Department E.B. Romantsova

Pat

**ADDITIONS AND CHANGES TO THE WORK PROGRAM FOR THE DISCIPLINE
MEDICAL GENETICS
SPECIALTY 31.05.01 MEDICAL CARE
FOR THE 2026-2027 ACADEMIC YEAR**

1. The tables in section 3.6. "Licensed and freely distributed software used in the educational process" shall be presented as follows:

List of software (commercial software products)

No. p/p	List of software (commercial software products)	Details of supporting documents
1.	MS operating system Windows 7 Pro	License number 48381779
2.	MS operating system Windows 10 Pro	CONTRACT No. UT-368 from September 21, 2021
3.	MS Office	License numbers: 43234783, 67810502, 67580703, 64399692, 62795141, 61350919
4.	Kaspersky Endpoint Security for Business - Standard Russian Edition . 50-99 Node 1-year Educational Renewal License	Agreement No. 7 AA dated 02/07/2025
5.	1C Accounting and 1C Salary	LICENSE AGREEMENT 612/L dated 02.02.2022 (additional licenses)
6.	1C: PROF University	LICENSE AGREEMENT No. KrTsB-004537 dated December 19, 2023
7.	1C: PROF Library	LICENSE AGREEMENT No. 2281 dated November 11, 2020
8.	Consultant Plus	Contract No. 41AA dated December 27, 2024
9.	Kontur.Tolk	Agreement No. K213753/24 dated August 13, 2024
10.	3KL e-learning environment (Russian Moodle)	Agreement No. 1362.5 dated November 20, 2024
11.	Astra Linux Common Edition	Agreement No. 142 A dated September 21, 2021
12.	Information system "Plans"	Agreement No. 2873-24 dated June 28, 2024
13.	1C: Document Management	Agreement No. 2191 dated 10/15/2020
14.	R7-Office	Agreement No. 2 KS dated 12/18/2020
15.	License for the "ROSA CHROME OS Workstation"	Agreement No. 88A dated 08/22/2024
16.	Alt Virtualization Server 10 (for secondary and higher vocational education)	Agreement No. 14AK dated September 27, 2024
17.	Dr.Web Desktop Security Suite Comprehensive Protection + Control Center	Agreement No. 8 dated October 21, 2024

	for 12 months.	
18.	Software "Schedule for educational institutions"	Agreement No. 82A dated July 30, 2024

List of freely distributed software

No . p/p	List of freely distributed software	Links to the license agreement
1.	Yandex Browser	Freely distributed License Agreement for the Use of Yandex Browser Software https://yandex.ru/legal/browser_agreement/
2.	Yandex.Telemost	Freely distributed License Agreement for the Use of Software https://yandex.ru/legal/telemost_mobile_agreement/
3.	Dr.Web CureIt !	Freely distributed License Agreement: https://st.drweb.com/static/new-www/files/license_CureIt_ru.pdf
4.	OpenOffice	Freely distributed License: http://www.gnu.org/copyleft/lesser.html
5.	LibreOffice	Freely distributed License: https://ru.libreoffice.org/about-us/license/
6.	VK Calls	Freely distributed https://vk.com/licence
7.	Kaspersky Free Antivirus	Freely distributed https://products.s.kaspersky-labs.com/homeuser/Kaspersky4Win2021/21.16.6.467/english-0.207.0/3830343439337c44454c7c4e554c4c/kis_eula_en-in.txt

2. The tables in section 3.5. “ Professional databases, information and reference systems, electronic educational resources ” shall be presented as follows:

Resource name	Resource Description	Access	Resource address
Electronic library systems			
Student Consultant. Medical University Electronic Library	For students and faculty of medical and pharmaceutical universities. Provides access to electronic versions of textbooks, teaching aids, and periodicals.	Remote access after registration under the university profile	https://www.studentlibrary.ru/
Reference and information system "	The MedBaseGeotar reference and information system is designed for practicing medical specialists,	Remote access after	https://mbasegeotar.ru/pages/index.html

MedBaseGeot ar "	researchers, teachers, postgraduate students, residents, senior students, and healthcare managers to quickly search, select, and read the medical literature they need for their work in a single data source.	registrati on under the universit y profile	
Electronic Library System "Bookup "	A large medical library is an information and educational platform for the shared use of electronic educational and methodological publications from medical universities in Russia and the CIS countries.	Remote access after registrati on under the universit y profile	https://www.books-up.ru/
Electronic Block System "Lan"	The Network Electronic Library of Medical Universities is an electronic database of educational and scientific works on medical topics, created for the purpose of implementing network forms of professional educational programs, open access to educational materials for partner universities.	Remote access after registrati on under the universit y profile	https://e.lanbook.com/
Scientific electronic library " CyberLeninka "	CyberLeninka is a scientific electronic library built on the Open Science paradigm. Its primary goals are the popularization of science and scientific activity, public oversight of the quality of scientific publications, the development of interdisciplinary research, a modern institution of scientific review, increasing the citation rate of Russian science, and building a knowledge infrastructure. It contains over 2.3 million scientific articles.	free access	https://cyberleninka.ru/
Human Biology Knowledge Base	Reference information on <u>physiology</u> , <u>cell biology</u> , <u>genetics</u> , <u>biochemistry</u> , <u>immunology</u> , <u>pathology</u> . (Resource <u>of the Institute of Molecular Genetics of the Russian Academy of Sciences</u> .)	free access	http://humbio.ru/
State Register of Medicines	The State Register of Medicines website contains information about medications: indications, contraindications, mechanism of action, side effects, dosages, and methods of administration.	free access	https://grls.rosminzdrav.ru/GR LS.aspx

Information systems			
Clinical Guidelines Index	A resource of the Russian Ministry of Health that contains clinical guidelines developed and approved by medical professional non-profit organizations of the Russian Federation, as well as methodological manuals, nomenclatures, and other reference materials.	Link to download the application	https://cr.minzdrav.gov.ru/#/
Federal Electronic Medical Library (FEMB)	The Federal Electronic Medical Library is part of the unified state information system in the field of healthcare as a reference system . The FEMB was created on the basis of the funds of the Central Scientific Medical Library named after I.M. Sechenov.	free access	https://femb.ru/
Russian State Library (RSL)	Collection size: approximately 3 million titles Period covered: from the 11th century to the present The Russian State Library's Electronic Library is a collection of electronic copies of valuable and frequently requested publications from the Russian State Library's collections, from external sources, as well as documents originally created in electronic form.	Registration on the website	https://www.rsl.ru/
Russian Medical Association	A professional online resource. Purpose: to promote effective professional activity among medical personnel. Contains the charter, personnel, structure, membership rules, and information about the Russian Medical Union.	free access	http://www.rmass.ru/
Web medicine	The website provides a directory of professional medical resources, including links to the most authoritative specialized websites, journals, societies, as well as useful documents and programs. It is intended for physicians, students, and staff of medical universities and research institutions.	free access	http://webmed.irkutsk.ru/

Databases			
World Health Organization	The site contains news, statistics on countries that are members of the World Health Organization, fact sheets, reports, WHO publications, and much more.	free access	http://www.who.int/ru/
Ministry of Science and Higher Education of the Russian Federation	The website of the Ministry of Science and Higher Education of the Russian Federation contains news, newsletters, reports, publications, and much more.	free access	http://www.minobrnauki.gov.ru
Ministry of Education of the Russian Federation	The website of the Ministry of Education of the Russian Federation contains news, newsletters, reports, publications, and much more.	free access	https://edu.gov.ru/
Polpred.com	Electronic library system Business media. Media review	free access	https://polpred.com/news
Bibliographic databases			
Database "Russian Medicine"	It is being created at the Central Scientific and Methodological Library and covers the entire collection since 1988. The database contains bibliographic descriptions of articles from Russian journals and collections, dissertations and their abstracts, as well as Russian and foreign books, collections of institute proceedings, conference materials, etc. Thematically, the database covers all areas of medicine and related areas of biology, biophysics, biochemistry, psychology, and	free access	https://rucml.ru/
PubMed	A text database of medical and biological publications in English. PubMed is an electronic search engine with free access to 30 million publications from 4,800 indexed medical journals. The database contains articles published from 1960 to the present, including information from MEDLINE, PreMEDLINE, and	free access	https://pubmed.ncbi.nlm.nih.gov/

	NLM. Each year, the portal is updated with more than 500,000 new papers.		
eLIBRARY.RU	A Russian information portal in science, technology, medicine, and education, containing abstracts and full texts of over 13 million scientific articles and publications. The eLIBRARY.RU platform offers electronic versions of over 2,000 Russian scientific and technical journals, including over 1,000 open-access journals.	Full functionality of the site is available after registration.	http://elibrary.ru/defaultx.asp
Electronic library of dissertations (RSL)	Currently, the Electronic Library of Dissertations of the Russian State Library contains more than 919,000 full texts of dissertations and abstracts.	free access	http://diss.rsl.ru/?menu=disscatalog/
Medline.ru	Medical and biological portal for specialists. Biomedical journal.	free access	https://journal.scbmt.ru/jour/index
Official Internet portal of legal information	The single official state information and legal resource in Russia	free access	http://pravo.gov.ru/

3. Add the following questions to the practical sections:

ma 5 . Chromosomal syndromes.

Question: Congenital malformations with chromosomal abnormalities.

Topic 6: Monogenic hereditary diseases with autosomal dominant inheritance.

Question: Monogenically inherited syndromes of congenital malformations.

Topic 12. Diseases with a hereditary predisposition (coronary heart disease, hypertension, diabetes, peptic ulcer, obesity, bronchial asthma).

Question: Multifactorial malformations of organs and organ systems.

Topic 13. Prevention of hereditary diseases. Medical and genetic counseling.

Question: prevention of congenital malformations.

Topic 14. Treatment of hereditary diseases. Gene therapy .

Question: Treatment of congenital malformations.