MINISTRY OF HEALTH OF UKRAINE

ODESSA NATIONAL MEDICAL UNIVERSITY

Department of medical biology and chemistry

APPROVED
Vice-rector for research and educational work,

_____ Eduard BURYACHKOVSKY

01 September 2024

CURRICULUM ON EDUCATIONAL DISCIPLINE

«MEDICAL GENETICS»

Level of higher education: second (master's)

branch of knowledge: 22 «Health Care»

specialty: 222 «Medicine»

educational and professional program: Medicine

2024

The curriculum is composed on the base of the educational and professional program «Medicine» for training specialists of the second (master's) level of higher education standard of in speciality 222 «Medicine» branch of knowledge 22 «Health care», approved by the Academic Council of ONMedU (minutes № 10 from 27 of June, 2024).

Developers:
Phd of Medical Sciences, Associate Professor Alla SHEVELENKOVA,
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Curriculum was discussed and approved at a methodic meeting of the Department of Medical Biology and Chemistry.
Minutes № 1, 26.08.2024.
Head of the department _____ Gennady STEPANOV

Agreed with the guarantor of EPP _____ Valeria MARICHEREDA

Curriculum was approvedon the meeting of the subject-cycle methodological comission for medical and biological disciplines ONMedU.
Minutes № 1, 27.08.2024.

Head of the committee for the medical and biological disciplines _____ Leonid GODLEVSKY

1. **Description of the discipline**

<table>
<tr><td>Name of indicators</td><td>Branch of knowledge, specialty, specialization, level of higher education</td><td colspan="2">Characteristics of the discipline</td></tr>
<tr><td rowspan="9">The total number of:
Credits – 3,0
Hours – 90
Content Modules – 5</td><td rowspan="9">branch of knowledge 22 «Health care»

speciality 222 «Medicine»

level of higher education second (master’s)</td><td colspan="2">Full-time education</td></tr>
<tr><td colspan="2">Selective</td></tr>
<tr><td>Year of study</td><td>IV</td></tr>
<tr><td>Semester</td><td>VII-VIII</td></tr>
<tr><td>Lectures</td><td>0</td></tr>
<tr><td>Practical classes</td><td>30 hours</td></tr>
<tr><td>Independent work</td><td>60 hours</td></tr>
<tr><td>Individual tasks</td><td>0</td></tr>
<tr><td>Forms of final control</td><td>Credit</td></tr>
</table>

2. The purpose and tasks of the discipline

The main **purpose** of discipline is the formation of knowledge and seminar skills in the field of etiology, pathogenesis, clinical manifestations, diagnostics and prevention of hereditary pathology.

The main **tasks** of studying the discipline are

1. To explain the etiology of chromosomal, single gene and multifactorial diseases.
2. To recognize the general manifestations of hereditary pathology, to diagnose the congenital morphogenetic variants, to use correctly the appropriate terminology in describing the clinical picture and phenotype of the patient.
3. To select the patients for cytogenetic, specific biochemical and molecular genetic diagnosis.
4. To make a preliminary conclusion on genetic risk in the most common single gene and chromosomal diseases
5. To identify high-risk groups for the development of hereditary diseases
6. To propose preventive measures of hereditary and congenital diseases.

The process of studying the discipline is aimed at forming elements of the following competencies:

Integral competence: The ability to solve typical and complex problems, including those of a research and innovation nature in the field of medicine. Ability to continue learning with a high degree of autonomy.

General competencies:

GC1 Ability to abstract thinking, analysis and synthesis.

GC2. Ability to learn and master modern knowledge

GC3. Ability to apply knowledge in seminar situations

GC4. Knowledge and understanding of the subject area and understanding of professional activity

GC11. Ability to search, process and analyze information from various sources

GC17. The desire to protect the environment

Special:

SC1. Ability to collect medical information about the patient and analyze clinical data

SC2. Ability to determine the necessary list of laboratory and instrumental studies and evaluate their results

SC24. Adherence to ethical principles when working with patients and laboratory animals

Program learning outcomes, the formation of which is facilitated by the educational discipline:

PLO2. Understanding and knowledge of basic and clinical biomedical sciences, at a level sufficient for solving professional tasks in the field of health care.

PLO4. To recognize and identify leading clinical symptoms and syndromes (according to list 1); according to standard methods, using preliminary data of the patient's history, data of the patient's examination, knowledge about the person, his organs and systems, establish a preliminary clinical diagnosis of the disease (according to list 2).

PLO21. Search for the necessary information in the professional literature and databases of other sources, analyze, evaluate and apply this information.

Upon completion of the study of the discipline "Medical Genetics" students must

To know:

- classification of mutations and mutagenic factors;
- concept, effects of genomic imprinting,
- lethal effects of mutations (significance in perinatal, early childhood and infant mortality, association with infertility, spontaneous abortion),
- classification of hereditary pathology,
- morphogenetic variants and their significance in the diagnosis of hereditary syndromes and inborn states.
- etiology, pathogenesis, classification of congenital defects, classification of teratogenic factors,
- principles and stages of clinical-genealogical examination.
- characteristic of pedigrees with different types of inheritance (autosomal-dominant, autosomal-recessive, X-linked, mitochondrial)
- general symptoms of chromosomal diseases,
- peculiarities of the clinical manifestations of individual syndromes: Down, Patau, Edwards, "cat cry", Prader-Willi, Angelman, Shereshevsky-Turner, Klinefelter, polysomy X, polysomy Y.
- indications for cytogenetic and molecular-cytogenetic diagnostics,
- medical genetic counseling in case of chromosomal diseases, the potentiality of prenatal diagnosis of chromosomal diseases,
- general questions of etiology and pathogenesis of single gene diseases, classification of single gene diseases,
- a general description of single gene diseases with different types of inheritance,
- clinical charachteristics, genetics, and diagnosis of Marfan syndrome, achondroplasia, congenital hypothyroidism, phenylketonuria, cystic fibrosis, congenital adrenal hyperplasia, fragile X- syndrome, Duchen-Becker muscular dystrophy,
- general characteristics and classification of inborn errors of metabolism,
- basic methods for diagnosis of suspected inborn errors of metabolism,
- indications for molecular-genetic and biochemical diagnostics,
- principles of selection of nosological forms for screening preclinical diagnostics,
- classification and general characteristics of multifactorial disorders,
- examples of multifactorial disorders with polygenic and monogenic predisposition,
- groups of genes that participate in oncogenesis,
- definition and examples of oncogenetic syndromes (OGS),

- levels of prophylaxis of hereditary diseases, ways of prevention,
- indications for medical genetic counseling (MGC), the purpose and objectives of the MGC,
- principles of preconceptional prevention,
- methods of prenatal diagnosis, basic principles and purpose of prenatal screening,
- deontological problems of MGC, prenatal diagnosis and screening genetic programs.

To be able:

- to conduct a conversation with the patient for collection of the genealogic history, to compose and analyze the pedigree,
- to be able to detect minor anomalies typical for chromosomal disorders, to reveal the symptoms of the most common single gene diseases (according to list 2) during the physical examination of the patient,
- to analyze the karyograms of patients with the most common chromosomal diseases, determine the type of structural or numerical chromosomal aberration.
- to identify indications for molecular-genetic, cytogenetic and biochemical diagnostics of hereditary diseases.
- to identify indications for medical genetic counseling.
- to calculate genetic risk in the most common hereditary diseases

3. Contents of the curriculum

Content Module 1. Heredity and pathology. The role of heredity in human pathology. Propaedeutic of hereditary disorders

Topic 1. Classification and etiology of hereditary disorders.

Subject and tasks of medical genetics. The role of medical-genetic knowledge in a practice of a physician. The place of medical genetics in the system of medical knowledge, the interrelations of medical genetics with other clinical and medical-prophylaxis disciplines. The growth of proportion of hereditary pathology in the structure of morbidity, mortality and invalidism of population. The relative increase of the proportion of hereditary diseases: population-genetic, environmental, social-economic and demographic aspects.

Classification of hereditary diseases. Single gene disorders. Disorders with hereditary predisposition. Chromosomal disorders.

Mutations as etiological factors. Numerical, structural chromosomal aberrations and gene mutations. Causes of mutations. Spontaneous and induced mutations. Physical, chemical and biological mutagens. Somatic and germ mutations. Disorders of somatic cells.

Epigenetic mutations. Chromosomal and genomic imprinting (the content, notion and effects).

Heredity and clinical manifestation. The clinical polymorphism and the modifying effect of genotype on the manifestation of pathological mutations. Genetic aspects of polymorphism of hereditary diseases.

Heredity and the outcome of diseases. Lethal effects of mutations (importance in the perinatal and infancy mortality, role in infertility, spontaneous miscarriage).

Topic 2. Semiotics of hereditary diseases.

Semiotics of hereditary diseases. Pleiotropic action of genes and multiple nature of the lesion in hereditary diseases. Primary and secondary pleiotropy in clinical manifestation of hereditary disorders. Peculiarities of manifestations of hereditary diseases. Hereditary diseases with late manifestation. Progressive course of the diseases. Affection of various organs and systems: polysystemic affection. Resistance to treatment in some forms.

Family as a subject of medical-genetic counseling: the need to family approach. Peculiarities of clinical examination of patients and their relatives, contributing to the diagnosis of congenital and hereditary diseases.

Morphogenetic variants of development (microanomalies, signs of dysembryogenesis), their genesis, postnatal modification. General and specific morphogenetic variants: value in the diagnostics of hereditary syndromes and congenital states.

Birth defects: primary and secondary. Isolated, systemic and multiple congenital birth defects (CBD). The concept of syndrome, association, deformation, dysplasia. Consistency of disorders character with the stages of ontogenesis: gameto-, embryo and fetopathy. Etiological heterogeneity of CBD. Hereditary, teratogenic, multifactorial malformations. Teratogenic factors.

Content Module 2. Chromosomal diseases.

Topic 3. General characteristics of chromosomal diseases. Cytogenetic methods.

Classification of chromosomal diseases. Age of parents and frequency of chromosomal diseases in children.

Pathogenesis of chromosomal diseases. Lethal effects of chromosomal and genomic mutations (spontaneous abortion, stillbirth, early infant mortality). Lethal effects of structural and numerical (genome) chromosomal aberrations (spontaneous abortions, stillbirth, early infant mortality).

General manifestations of chromosomal disorders. The peculiarities of pregnancy course in chromosomal disorders of a fetus. Birth defects, the involvement of different systems in the pathological process, craniofacial dysmorphy, delay of psychomotor development, mental retardation, endocrinopathy. Progressive course.

Methods of diagnosis of chromosomal disorders. Cytogenetic methods.

Indications for cytogenetic diagnostics. Classification of cytogenetic method.

Medical-genetic counseling. Prenatal diagnosis of chromosomal diseases.

Topic 4. Chromosomal disorders caused by structural and numerical aberrations of autosomes.

Peculiarities of clinical manifestations of Down syndrome. Peculiarities of clinical manifestations of syndromes: Patau, Edwards, "cri-du-chat" Angelman, Prader-Willy. Population frequency. Possibilities of therapy and rehabilitation

Topic 5. Chromosomal disorders caused by numerical aberrations of sex chromosomes.

Peculiarities of clinical manifestations of syndromes: Shereshevsky-Turner, Klinefelter, polysomy X, polysomy Y. Population frequency. Possibilities of therapy and rehabilitation

Content Module 3. Single gene diseases.

Topic 6. General characteristics and classifications of single gene disorders. Genealogical method.

General questions of etiology and pathogenesis of single gene disorders. Clinical importance of incomplete penetrance, variable expressivity and modifying environmental factors in clinical polymorfism of etiologically same forms of hereditary pathology. Genetic heterogeneity of clinically similar forms of disease. Aspects of heterogeneity: allelic and locus heterogeneity (clinical examples). Notion of geno-, pheno- and normocopy.

Common and orphan forms. Prevalence among the various contingents.

Classifications of single gene diseases: etiological (genetic), organ-system, pathogenetic.

Genealogical method. Stages of clinical-genealogical examination. Basic notions: pedigree, proband (propositus), pedigree legend, symbols. Methods of genealogical information gathering and its peculiarities in various diseases.

The role of clinical-genealogical method in clinical practice for determination of the nature of the disease, estimation of clinical manifestations, differential diagnostics of hereditary forms of pathology, the study of genetic heterogeneity of disease, estimation of the risk of new cases in the family, the prognosis of the disease and life.

Criteria for different types of inheritance: autosome-dominant, autosome-recessive, X-linked dominant, X-linked recessive, holandric, mitochondrial. The characteristics of the pedigrees,

the sex ratio, segregation of pathological signs in the family. The dependence of the pedigree nature on the frequency of genes in the population. Recessive pathology and consanguinity. The notion of "sporadic case", the possible causes of "sporadic cases" in the family, the *de novo* mutation. The phenomenon of anticipation.

Topic 7. Single gene disorders with autosomal-dominant and X-linked modes of inheritance.

Single gene disorders with autosomal-dominant mode of inheritance. General characteristics. Marfan syndrome, achondroplasia. Phenotypes and genotypes of patients. Principles of diagnosis. Medical-genetic counselling.

Topic 8. Single gene disorders with X-linked mode of inheritance.

X-linked disorders. General characteristics. Fragile-X syndrome, Duchenne-Becker muscular dystrophy. Phenotypes and genotypes of patients. Principles of diagnosis. Medical-genetic counselling.

Topic 9. Single gene disorders with autosomal-recessive mode of inheritance. **Inborn errors of metabolism.**

Single gene disorders with autosomal-recessive mode of inheritance. General characteristics. The characteristics of the pedigrees with autosomal recessive inheritance.

Inborn errors of metabolism: principles of classification, pathogenesis, principles of diagnosis and treatment.

Clinics and genetics of some single gene disorders (phenylketonuria, congenital hypothyroidism, cystic fibrosis, adrenal hyperplasia). Frequency in population, clinical forms and variants, types of mutations, pathogenesis, typical clinical manifestations, paraclinical and laboratory methods of diagnostics, symptomatic and pathogenetic treatment, prognosis, rehabilitation, social adaptation. Etiologic treatment. Genetic engineering approaches in the treatment of hereditary diseases. Indications for molecular-genetic analysis.

Molecular-genetic and biochemical methods of diagnosis of single gene disorders.

Screening program of mass diagnostics of hereditary diseases and heterozygous states.

Mitochondrial inheritance. General characteristics of mitochondrial pathology. Mitochondrial disorders. Examples, general principles of diagnosis and treatment of mitochondrial pathology.

Content Module 4. Diseases with hereditary predisposition.

Topic 10. General characteristics and classification of multifactorial diseases.

The role of hereditary and environmental factors in the origin of common pathology of noninfectious etiology. The concept of predisposition. Genetic polymorphism of populations. The interaction of genetic predisposition and specific environmental conditions in the development of diseases.

General characteristics of multifactorial diseases: high frequency in the population, the nature of gender-age differences, peculiarities of the prevalence of susceptibility genes and disease in families.

Classification of multifactorial diseases: disease with polygenic and monogenic predisposition.

Polygenic predisposition as a result of nonallelic genes interaction. Genetics of multifactorial diseases: the terminology, notion and content. Degree of risk dependence on the degree of relationship with the propositus, the severity of his condition, proband's sex, population frequency, occupation and living conditions. Tables of empiric risk. Markers of predisposition. High risk factors.

Congenital defects of multifactorial origin. Examples.

Monogenic predisposition: ecogenetic pathology, pharmacogenetic reactions. Genetically based peculiarities of biotransformation of the drugs.

Oncogenetic syndromes (OGS). Definition, examples. Groups of genes that participate in oncogenesis. Hereditarily determined forms of neoplasia.

Content Module 5. Diagnosis and prevention of hereditary diseases. Medical-genetic counseling.

Topic 11. Methods of laboratory diagnosis of hereditary disorders

Indications for cytogenetic diagnostics. Classification of cytogenetic methods. Modern methods of karyotyping, molecular-cytogenetic methods (fluorescence *in situ* hybridization), sex chromatin detection. The value of the cytogenetic method in clinical practice: the diagnosis of chromosomal diseases, the diagnosis of Mendelian diseases associated with chromosome instability, the diagnosis of some cancer diseases and forms of leukemia, assessment of mutagenic effects of drugs, monitoring of the effects of environmental factors.

Indications for molecular-genetic analysis. Characteristics of the main methodological approaches (DNA extraction, restriction of DNA, blot hybridization, sequencing). The method of PCR (polymerase chain reaction) method for RFLP (restriction fragment length polymorphism).

The new methods of identifying mutations - method of DNA – micro arrays.

Indications for biochemical diagnostics. Stages of biochemical diagnosis: selective screening, confirmatory diagnosis. Levels of biochemical diagnostics: primary gene product, cell level, metabolites in biological fluids.

Screening diagnostics: qualitative and quantitative methods. List of the basic methods and their brief description (qualitative tests in urine, paper and thin-layer chromatography of amino acids and carbohydrates in the urine and blood, electrophoresis, Gatri's microbiological inhibitory test, fluorometry, etc.). Screening program of mass diagnostics of hereditary diseases and heterozygous states.

The confirmative diagnostics. Quantitative determination of enzymes and metabolites. Modern methods: automatic analysis of amino acids, liquid and gas chromatography, mass spectrometry, and radio immunochemical and immunoenzymologic assays.

Topic 12. Levels and ways of prevention of hereditary diseases.

Types of prevention of hereditary diseases: primary, secondary and tertiary prevention. Levels of prevention: pregametic, prezigotic, prenatal and postnatal. Ways of carrying out of preventive measures: influence on the penetrance and expressivity; elimination of the embryo and fetus; family and fertility planning, protection of the environment. The forms of prophylaxis: medical-genetic counseling, prenatal diagnostics, mass screening program; "genetic" clinical dispanserization (registers), the environment protection and control of mutagenic environmental factors.

Topic 13. Medical-genetic counseling. Credit

Medical-genetic counseling (MGC) as a type of specialized medical care. The task of the MGC and the indications for the direction of the patients and their families at MGC. Prospective and retrospective counseling. Genetic risk, risk degrees. The notion of theoretical and empirical risk. Principles of genetic risk assessing for monogenic, chromosomal and multifactorial pathology. The methodic of the MGC. Calculations of genetic risk communication information to patients, aid to the family in their decision. Deontological and ethical questions of MGC. Cooperation of physiciansin the MGC. Organization of medical genetic aid in Ukraine.

Prenatal diagnosis as a method of prevention. Common indications for prenatal diagnostics. Noninvasive methods of prenatal diagnosis: ultrasonography, detection of the biochemical markers in the maternal blood serum, NIPT.

Invasive methods. Methods for obtaining a fetal material: biopsy of chorion and placenta, amnio- and cordocentesis. Indications, terms, contraindications and possible complications.

Deontological and ethical questions that arise during prenatal diagnosis.

Screening programs. Principles of selection of nosological forms for screening preclinical diagnostics. Characteristics of the main diagnostic programs of phenylketonuria, congenital

hypothyroidism, congenital adrenal hyperplasia, cystic fibrosis. Diagnostics of heterozygous states in groups of high genetic risk. Deontological questions of screening programs.

4. Structure of the discipline "Medical genetics"

Topics	Hours			
	Total	Including		
		L	S.c.	ISW
Content Module 1. Heredity and pathology. The role of heredity in human pathology				
Topic 1. Classification and etiology of hereditary disease	9	0	4	5
Topic 2. Semiotics of hereditary diseases.	6	0	2	4
Totally for content module	15	0	6	9
Content Module 2. Chromosomal diseases.				
Topic 3. General characteristics of chromosomal diseases. Cytogenetic methods of diagnosis	6	0	2	4
Topic 4. Chromosomal disorders caused by structural and numerical aberrations of autosomes.	7	0	2	5
Topic 5. Chromosomal disorders caused by numerical aberrations of sex chromosomes.	6	0	2	4
Totally for content module	19		6	13
Content Module 3. Single gene disorders.				
Topic 6. General characteristics of single gene disorders. Genealogical method	6	0	2	4
Topic 7. Single gene disorders with autosomal-dominant mode of inheritance.	7	0	2	5
Topic 8. Single gene disorders with X-linked mode of inheritance.	6	0	2	4
Topic 9. Single gene disorders with autosomal-recessive mode of inheritance. Inborn errors of metabolism.	7	0	2	5
Totally for content module	26	0	8	18
Content Module 4. Diseases with hereditary predisposition.				
Topic 10. General characteristics and classification of multifactorial diseases.	7	0	2	5
Totally for content module	7	0	2	5
Content Module 5. **Diagnosis and prevention of hereditary diseases. Medical-genetic counseling.**				
Topic 11. Methods of laboratory diagnosis of hereditary disorders	7	0	2	5
Topic 12. Levels and ways of prophylaxis of the hereditary diseases.	7	0	2	5
Topic 13. Medical-genetic counseling. Credit	9	0	4	5
Totally for content module	23	0	8	15
Total	90	0	30	60

5. Topics of the lectures / seminar / seminar / laboratory classes

5.1. Topics of the lectures

Lectures are not planned

5.2. Topics of the practical classes

№	Topic	Hours
1	Classification and etiology of hereditary disease	4
2	Semiotics of hereditary diseases	2
3	General characteristics of chromosomal diseases. Cytogenetic methods	2
4	Chromosomal disorders caused by structural and numerical aberrations of autosomes.	2
5	Chromosomal disorders caused by numerical aberrations of sex chromosomes.	2
6	***General characteristics of single gene disorders. Genealogical method***	2
7	Single gene disorders with autosomal-dominant mode of inheritance	2
8	Single gene disorders with X-linked mode of inheritance	2
9	Single gene disorders with autosomal-recessive mode of inheritance. Inborn errors of metabolism.	2
10	General characteristics and classification of multifactorial diseases	2
11	Methods of laboratory diagnosis of hereditary disorders	2
12	Levels and ways of prophylaxis of the hereditary diseases.	2
13	Medical-genetic counseling/ Credit	4
	Total	30

5.3. Topics of laboratory classes

Laboratory classes are not planned.

7. Independent students work

№	Topic	Hours
1	Preparing for practical class 1	5
2	Preparing for practical class 2	4
3	Preparing for practical class 3	4
4	Preparing for practical class 4	5
5	Preparing for practical class 5	4
6	Preparing for practical class 6	4
7	Preparing for practical class 7	5
8	Preparing for practical class 8	4
9	Preparing for practical class 9	5
10	Preparing for practical class 10	5
11	Preparing for practical class 11	5
12	Preparing for practical class 12	5
13	Preparing for practical class 13	5
	Total	**60**

7. Teaching methods

Practical classes: conversation, explanations, case studies, acquiring knowledge of the etiology, pathogenesis of hereditary diseases, mastering the skills of analyzing the phenotypes of patients in order to determine the clinical manifestations of hereditary pathology, evaluating the

diagnostic and prognostic value of the detected symptoms, morphogenetic variants (microanomalies of development), practicing the skills of compiling and analyzing pedigrees, acquiring knowledge and mastering skills in the diagnosis of the most common forms of hereditary pathology.

Understanding the goals, knowledge of the methods and possibilities of medical genetic counseling, prenatal diagnosis and screening programs.

Independent work: independent work with the textbook, independent work with the data base of MCQ tasks, independent case studies.

8. Forms of control and evaluation methods (including criteria for evaluating learning outcomes)

Current control is carried out on the basis of control of theoretical knowledge, seminar skills and abilities: oral interview, testing, evaluation of the performance of seminar works (analysis of phenotypes of patients with hereditary diseases, analysis of pedigrees), solving of situational tasks, evaluation of activity in class.

Final control: credit.

The structure of the current assessment in the seminar lesson:

1. Assessment of theoretical knowledge on the topic of the lesson:
- methods: recitation, interview, case studies;
- maximum grade - 5, minimum grade - 3, unsatisfactory grade - 2.
2. Assessment of seminar skills and abilities on the topic of the lesson:
- Case studies, methods: assessing the accuracy of seminar skills conducting
- maximum score - 5, minimum score - 3, unsatisfactory score - 2;

Criteria for current assessment in the practical lesson:

Exellent «5»	The student is fluent in the material, takes an active part in the discussion and case studies, confidently demonstrates seminar skills during performing and interpreting of seminar work on the topic of the lesson, expresses his opinion on the topic of the lesson.
Good «4»	The student is well versed in the material, participates in the discussion and case studies, demonstrates seminar skills while performing seminar work with some mistakes, expresses his opinion on the topic of the lesson.
Satisfactorily «3»	The student does not have enough knowledge in theoretical material, insecurely participates in the discussion and case studies, performs seminar work with significant errors.
Unsatisfactorily «2»	The student does not have knowledge in theoretical material, does not participate in the discussion and case studies, does not demonstrate seminar skills during the seminar work on the topic of the lesson.

Credit is given to the applicant who completed all tasks of the work program of the academic discipline, took an active part in practical classes, completed and defended an individual assignment and has an average current grade of at least 3.0 and has no academic debt.

Assessment is carried out: at the last lesson before the beginning of the examination session - with the tape system of learning, at the last lesson - with the cyclical system of learning. The credit score is the arithmetic mean of all components according to the traditional four-point scale and has a value that is rounded according to the statistics method with two decimal places after the decimal point.

9. Distribution of points obtained by applicants of higher education

The average score for the discipline is translated into a national score and converted into points on a multi-point scale (200-point scale).

Converting of the traditional grade from the discipline to 200-point is performed by the information and computer center of the university program "Contingent" by the formula: average grade point of success (current /in discipline) x 40

National grade for the discipline	Total grades for the discipline
Exellent «5»	**185-200**
Good «4»	**151-184**
Satisfactorily «3»	**120-150**
Unsatisfactorily «2»	**Less then 120**

A multi-point scale (200-point scale) characterizes the actual success of each applicant in learning the educational component. The conversion of the traditional grade (average score for the academic discipline) into a 200-point grade is performed by the information and technical department of the University.

According to the obtained points on a 200-point scale, the achievements of the applicants are evaluated according to the ECTS rating scale. Further ranking according to the ECTS rating scale allows you to evaluate the achievements of students from the educational component who are studying in the same course of the same specialty, according to the points they received.

The ECTS scale is a relative-comparative rating, which establishes the applicant's belonging to the group of better or worse among the reference group of fellow students (faculty, specialty). An "A" grade on the ECTS scale cannot be equal to an "excellent" grade, a "B" grade to a "good" grade, etc. When converting from a multi-point scale, the limits of grades "A", "B", "C", "D", "E" according to the ECTS scale do not coincide with the limits of grades "5", "4", "3" according to the traditional scale. Acquirers who have received grades of "FX" and "F" ("2") are not included in the list of ranked acquirers. The grade "FX" is awarded to students who have obtained the minimum number of points for the current learning activity, but who have not passed the final examination. A grade of "F" is assigned to students who have attended all classes in the discipline, but have not achieved a grade point average (3.00) for the current academic activity and are not admitted to the final examination.

Applicants studying in one course (one specialty) are ranked on the ECTS scale as follows, based on the number of points scored in the discipline:

ECTS Point	Statistics indicator
«A»	The best 10 % of the students
«B»	Next 25 % students
«C»	Next 30 % students
«D»	Next 25 % students
«E»	The last 10 % students

10. Methodological support:

• Curriculum of the discipline;
• Syllabus of the discipline;
• Power point presentations;
• Sets of the photokaryograms, pedigrees and phenotypes of patients with hereditary disorders;
• Methodical instructions for students for seminar classes and independent work;
• Case tasks;
• Electronic data bank of MCQ tasks.

11. Control questions on medical genetics

1. Subject and tasks of medical genetics. The role of genetics in medicine.
2. Classification of mutations. Spontaneous and induced, somatic and germ mutations. Numerical and structural chromosomal aberrations, gene mutations. Balanced and unbalanced chromosomal aberrations.
3. Definition and classification of hereditary diseases.
4. Semiotics of hereditary diseases. Peculiarities of clinical manifestations of congenital and hereditary diseases.
5. Peculiarities of examination of the patient and his family: congenital defects, minor anomalies of the development.
6. Syndromologic approach in the diagnosis of congenital and hereditary diseases.
7. Clinical-genealogic method. Methodic of pedigree composition and analysis. Modes of inheritance.
8. Chromosomal diseases. Etiology and classification.
9. Pathogenesis of chromosomal diseases.
10. General symptoms of chromosomal diseases.
11. Clinical and genetic characteristics of Patau, Edwards and Down syndromes.
12. Clinical and genetic characteristics of Turner syndrome, polysomy in sex chromosomes in males and females.
13. Clinical and genetic characteristics of syndromes with partial aneuploidy. Cri-du-chat syndrome.
14. Clinical and genetic characteristics microcytogenetic syndromes. Angelman and Prader – Willi syndromes.
15. Genomic imprinting. Definition. Genomic imprinting diseases. Etiology, pathogenesis, clinical forms. Angelman and Prader –Willi syndromes.
16. Medical-genetic counseling in chromosomal diseases. Factors of increased risk for children with chromosomal diseases.
17. Cytogenetic and molecular cytogenetic methods. Indications for cytogenetic studies.
18. Monogenic (single gene) diseases. Etiology and classification.
19. General characteristics of autosomal-dominant diseases. The clinic, genetics and diagnosis of Marfan syndrome, achondroplasia.
20. General characteristics of autosomal-recessive diseases. The clinical manifestations, genetics and diagnostics of congenital hypothyroidism, phenylketonuria.
21. General symptoms of inborn errors of metabolism. Principles of diagnosis. Classification of inborn errors of metabolism.
22. General characteristics of X-linked recessive diseases. The clinical manifestations, genetics and diagnostics of Duchenne-Becker muscular dystrophy, fragile X syndrome.
23. Biochemical methods. Indications for biochemical diagnostics.
24. Indications for biochemical mass screening of newborns.
25. Molecular genetic methods. Indications and possibilities of this method.
26. Prevention of congenital and hereditary diseases. Types of prevention.

27. Preconception prevention.
28. Medical-genetic counseling (MGC). The purpose and objectives of MGC, indications to the MGC.
29. Prenatal diagnosis (PD). Classification of the methods: invasive and noninvasive.
30. Prenatal screening of the first and second trimester of pregnancy.

Indicative list of practical skills

1. To analyze the phenotype of patient with hereditary disorder, to determine a preliminary diagnosis.
2. To make a plan of genetic examination of the patient.
3. To calculate the genetic risk of hereditary disease in the family.
4. To make a plan of prenatal diagnosis of hereditary diseases.
5. To determine the tactics of curation of a patient with Down, Klinefelter, Shereshevsky-Turner syndromes, phenylketonuria and other most common genetic disorders.
6. To analyze the karyotype of the patient and determine the diagnosis of chromosomal disorder.
7. To compose and analyze the pedigree and identify the mode of inheritance.

12. List of recommended materials

Main literature

1. Methodical recommendations on medical genetics

Additional literature:

1. Genetics in medicine. - 7th edition/Robert L/Nussbaum, Roderick R. McInnes, Huntington F. Willard. – 2007 – 585 p.
2. Emery's Elements of medical genetics. 15th ed. / Peter Turnpenny, Sian Ellard. – Elsevier, 2017. – 400 pp.
3. Lynn B. Jorde, John C. Carey, Michael J. Bamshad. Medical genetics. 5th ed. Elsevier, 2016. 356 pp.
4. Vogel and Motulsky's human genetics. Problems and approaches / M. R. Speicher, S. E. Antonarakis, F. G. Motulsky. 4th addition. – Springer, 2010. – 981 pp.
5. Young Ian.D. Medical genetics. -2nd ed. -Oxford university press, 2010. - 304 p.
6. Diseases of the fetus and newborn. Pathology, radiology and genetics. G.B.Reed, A.E. Claireaux and A.D.Bain., - Great Britain, - 1989, 812 p.
7. Human molecular genetics. Tom Strachan, Andrew P.Read. – 4th edition - Bios Scientific Publisher, 2010, 680 p.
8. Smith recognizable patterns of human malformation. Seventh edition. John M. Graham, - USA, - 2013, 976 p.
9. R. Wiedemann, K.-R. Gross, H.Dibberin ,Atlas of characteristic syndromes. A Visual Aid to Diagnosis. Second edition. -London, 1986,412 p.

13. Information resources:

https://ghr.nlm.nih.gov National librery of medicine, genetics
https://www.orpha.net The portal for rare diseases and orphan drugs
https://rarediseases.org National Organization for Rare Disorders
http://omim.org/OMIM (Online Mendelian Inheritance in Man) – An Online Catalog of Human Genes andGenetic Disorder