MINISTRY OF HEALTH OF UKRAINE
ODESSA NATIONAL MEDICAL UNIVERSITY
Department of clinical immunology, genetics and medical biology

APPROVED
A.i. vice-rector for research and educational work
Professor _____ I.P. Shmakova
"01" 09 2021

CURRICULUM ON EDUCATIONAL DISCIPLINE
"MEDICAL GENETICS"

Level of higher education: second (master's)

Branch of knowledge: 22 «Health care»

Speciality: 222«Medicine»

Educational-professional program: Medicine

Odessa 2021 - 2022

The curriculum is composed on the base of the educational-qualification program "Medicine" for training specialists of the second (master's) level of higher education standard of higher education of Ukraine in specialty 222 "Medicine" in the field of knowledge 22 "Health care", approved by the Academic Council of ONMedU, from 04.06.2020, protocol №11.

Developers: Phd of Medical Sciences, Associate Professor Shevelenkova A.V., Phd of Medical Sciences, Associate Professor Chesnokova M.M.

Program was discussed and approved on the methodic meeting of the department at 27.08.2021. Protocol № 1

Head of the department
Professor _____ **Goncharuk S. F.**

Program
was approved on the meeting of the cycle methodical committee of the medical and biological disciplines on 27.08. 2021. Protocol № 1

Head of the committee of the medical and biological disciplines
Professor _____ **Appelhans O. L.**

Program was approved on the Central methodical committee meeting of ONMedU
Protocol № 1 on 30.08. 2021

1. Description of the discipline

Name of indicators	Characteristics of the discipline	
	Full-time education	
The total number of: Credits - 1.0 Hours - 30 Content sections - 6	Elective discipline	
	Year of preparation	4
	Semester	VII-VIII
	Lectures	0
	Practical classes	20 hours
	Independent work	10 hours
	Individual tasks	0
	Form of final control	Differential credit

2. The purpose and tasks of the discipline

The main **purpose** of discipline is the formation of knowledge and practical 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 competencies of magister:***

- Ability to solve complex problems and problems in a certain field professional activity or in the process of learning that involves conducting research and/or implementing innovations and characterized by complexity and uncertainty of conditions and requirements.

- Genera competencies:

GC1. Ability to abstract thinking, analysis and synthesis.

GC2. Knowledge and understanding of the subject area. and understanding of professional activity.

GC 4. Ability to learn and master modern knowledge, use information and communication technologies; ability to search, process and analyze information from various sources.

GC9 Ability to act on the basis of ethical considerations, socially responsible and conscious.

Special:

SC1 Communication skills and clinical examination of the patient

SC2 Ability to determine the list of required clinical laboratory and instrumental studies and evaluate their results

SC9 Ability to provide advice on family planning, to determine the tactics of physiological pregnancy, physiological childbirth and the postpartum period.

Learning outcomes for discipline:

"Medical genetics" as an educational discipline sets the foundation for the further formation of the following **program educational outcomes**:

PEO 1 To have communication skills and skills of clinical examination of a patient. To collect data on patient complaints, medical history, life history.

PEO 3 Highlight the leading clinical symptom or syndrome (according to list 1). Establish a preliminary diagnosis, make a differential diagnosis and determine the clinical diagnosis of the disease (according to list 2).

PEO 9 To provide consultations on family planning, to determine the tactics of physiological pregnancy, physiological childbirth and the postpartum period.

PEO 16 Assess the impact of the environment, socio-economic and biological determinants on the health of the individual, family, population.

PEO 18 Adhere to the requirements of ethics, bioethics and deontology in their professional activities.

Upon completion of the study of the discipline "Medical Genetics" students must **know**:

- frequency of congenital and hereditary pathology in different periods of ontogenesis
- 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, cystic fibrosis, congenital hypothyroidism, phenylketonuria, congenital adrenal hyperplasia, fragile X- syndrome, Duchen-Becker muscular dystrophy,
- general description of mitochondrial pathology, examples of mitochondrial diseases (MERRF syndrome and MELAS syndrome);
- 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,
- PCR method, as the basic method of molecular diagnostics,
- principles of selection of nosological forms for screening preclinical diagnostics,

- the concept of predisposition, examples of genes of predisposition, genetic polymorphism of populations,
- general description of multifactorial diseases, principles of classification,
- examples of multifactorial diseases with a monogenic and polygenic predisposition,
- general characteristics of oncogenetic, pharmacogenetic and ecogenetic syndromes,
- 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,
- general principles of treatment of hereditary diseases, rehabilitation and social adaptation of patients.
- 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 chapter 1. Heredity and pathology. The role of heredity in human pathology

Topic 1. Subject and tasks of medical genetics. **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 presentation. 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).

Content chapter 2. Propaedeutic of hereditary disorders. Clinical-genealogical method.

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, micro signs, 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.

Family as an object of medical genetic observation: the need for a family approach.

Topic 3. Clinical-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 types of pathology.

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: autosomal-dominant, autosomal-recessive, X-linked dominant,X-linked recessive, holandric, mitochondrial. The characteristics of the pedigrees, the sex ratio, segregation of pathological traits 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.

Content chapter3. Chromosomal diseases.

Topic 4. General characteristics of chromosomal diseases. Down syndrome.

Notion of karyotype. Etiology and cytogenetics of chromosomal diseases. Classification of chromosomal diseases. Structural and numerical (genome) chromosomal aberrations. Partial trisomy and monosomy. Complete and mosaic forms. Uniparental disomy. Chromosomal imprinting. Parental age and the frequency of chromosomal diseases in children.

Pathogenesis of chromosomal diseases.

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.

Peculiarities of clinical manifestations of Down syndrome.

Methods of diagnosis of chromosomal disorders. Medical-genetic counseling. Prenatal diagnosis of chromosomal diseases.

Topic 5. Clinical manifestation of main forms of chromosomal diseases.

Peculiarities of clinical manifestations of syndromes: Patau, Edwards, "cri-du-chat" Angelman, Prader-Willy, Shereshevsky-Turner, Klinefelter, polysomy X, polysomy Y. Population frequency. Possibilities of therapy and rehabilitation.

Content chapter 4. Single gene disorders.

Topic 6. Single gene disorders with autosomal-dominant and X-linked modes of inheritance.

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.

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

Single gene disorders with autosomal-dominant mode of inheritance. General characteristics. Marfan syndrome, achondroplasia.

X-linked disorders. General characteristics. Fragile-X syndrome. Duchenne-Becker muscular dystrophy.

Topic 7. Single gene disorders with autosomal-recessive mode of inheritance. **Inborn errors of metabolism.**

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

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

Clinics and genetics of some single gene disorders (phenylketonuria, congenital adrenal hyperplasia, cystic fibrosis, congenital hypothyroidism). 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 and stem cell therapy in the treatment of hereditary diseases.

Mitochondrial inheritance. General characteristics of mitochondrial diseases. Classification of mitochondrial diseases. Examples, general principle of diagnostics and treatment of mitochondrial pathology.

Content chapter 5. Diagnosis and prevention of hereditary diseases. Medical-genetic counseling and prenatal diagnosis

Topic 8. Laboratory diagnosis of hereditary disorders

Indications for cytogenetic diagnostics. Classification of cytogenetic method. 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. The new methods of mutations identifying (DNA micro arrays).

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

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

Topic 9. Level and ways of prevention of hereditary diseases. Medical-genetic counseling.

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.

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 level 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 program. 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.

Content chapter 6. 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 multifactorialdiseases: high frequency in the population, the nature of sex and age differences, peculiarities of the prevalence of susceptibility genes and disease in families. Principles of genetic risk calculation.

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

Polygenic predisposition as a result of non-allelic genes interaction. 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 carcinogenesis. Hereditarily determined forms of neoplasia.

4. Structure of the discipline “Medical genetics”

Тема	Hours			
	Total	Including		
		L	P..c.	ISW
Content chapter 1. Heredity and pathology. The role of heredity in human pathology				
Topic 1. Classification and etiology of hereditary disease	2	0	2	
Content chapter 2. Propaedeutic of hereditary disorders. Clinical-genealogical method.				
Topic 2. Semiotics of hereditary diseases.	2	0	2	

Topic 3. Clinical-genealogical method.	2	0	2	
Content chapter 3. Chromosomal diseases.				
Topic 4. General characteristics of chromosomal diseases. Down syndrome.	2	0	2	
Topic 5. Clinics of the main forms of chromosomal diseases.	2	0	2	
Content chapter 4. Single gene disorders.				
Topic 6. Single gene disorders with autosomal-dominant and X-linked modes of inheritance.	2	0	2	
Topic 7. Single gene disorders with autosomal-recessive mode of inheritance. Inborn errors of metabolism.	4	0	2	2
Content chapter 5. Diagnostics and prevention of the hereditary diseases. Medical-genetic counseling and prenatal diagnosis..				
Topic 8. Laboratory diagnostics of hereditary disorders	2	0	2	
Topic 9. Levels and ways of prophylaxis of the hereditary diseases. Medical-genetic counseling.	2	0	2	
Content chapter 6. Diseases with hereditary predisposition.				
Topic 10 General characteristics and classification of multifactorial disorders.	4	0	0	4
Differential credit	6		2	4
Total	30	0	20	10

Auditory work— 66,6%, ISW – 33,4%

5. Topics of the lectures

Not provided by the curriculum

6. Topics of the practical classes

№	Topic	Hours
1	Classification and etiology of hereditary disease	2
2	Semiotics of hereditary diseases. Congenital defects	2
3	Clinical-genealogical method	2
4	General characteristics of chromosomal diseases. Down syndrome	2
5	Clinical characteristic of most common forms of the chromosomal diseases	2
6	Single gene disorders with autosomal-dominant and X-linked modes of inheritance.	2
7	Single gene disorders with autosomal-recessive mode of inheritance. Inborn errors of metabolism.	2
8	Laboratory diagnostics of hereditary disorders	2
9	Levels and ways of prophylaxis of the hereditary diseases. Medical-genetic counseling.	2
10	Differential credit	2
	Total	20

7. Independent students work

№	Topic	Hours
1.	***Self-elaboration*** **on the themes that are not included into the plan of classroom work**	
1.1	Lysosomal storage disorders. Mitochondrial disorders	2

1.2	Multifactorial disorders. Pharmacogenetic, ecogenetic and oncogenetic syndromes.	4
2.	Preparing for differential credit	4
	Total	10

8. **Individual research work:**

1. Review of the literature with writing of an essays.
2. Participation in medical genetic counseling with the report about clinical case.
3. Participation in student conferences with the report on medical genetics.
4. Participation in the Olympiad on medical genetics.
5. Work in cytogenetic or molecular-genetic laboratory

9. Teaching methods

Practical classes: conversation, explanations, case studies, practical work on phenotype analysis, pedigree composing, obtaining of skills on diagnosis of most common hereditary pathology.

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

10. Methods of control and evaluation criteria of educational outcomes

Current control is carried out on the basis of control of theoretical knowledge, practical skills and abilities: recitation, testing, assessment of practical skills conducting, case studies, assessment of activity in the classroom.

Final control: differential credit.

The structure of the current assessment in the practical lesson:

Assessment of current performance is based on the results:

- interviews in a practical lesson;
- solving case problems;
- analysis of phenotypes of patients with hereditary diseases;
- analysis of pedigrees with different types of inheritance;
- calculation of genetic risk;
- composing a plan of diagnosis and prenatal diagnosis.

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 practical skills and abilities on the topic of the lesson:

- Case studies, methods: assessing the accuracy of practical skills conducting (- analysis of phenotyp patients with hereditary diseases; analysis of pedigrees with different types of inheritance; calculation of ge risk; composing a plan of diagnosis and prenatal diagnosis)
- maximum score - 5, minimum score - 3, unsatisfactory score - 2;

Criteria for current assessment in the practical lesson:

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

«2»	The student does not have knowledge in theoretical material, does not participate in the discussion and case studies, does not demonstrate practical skills during the practical work on the topic of the lesson.

Final control of the discipline - **differential test**. The differential test is set at the last lesson of the discipline with the obligatory performance by the student of all types of work provided by the working curriculum and evaluated for the current educational activity on average not less than 3.00. Differential credit is made in the form of a written answer from the student. Each task includes 40 MCQ tasks, pedigree analysis, phenotype of a patient with hereditary pathology, situational problem and 2 theoretical questions. Differential credit is assessed on a 4-point scale.

Criteria for assessing the educational outcomes of students at the differential test:

«5»	is given to the student who systematically worked during a semester, showed during examination various and deep knowledge of a program material, is able to successfully carry out tasks which are provided by the program, has mastered the maintenance of the basic and additional literature, has understood interrelation of separate sections of discipline. importance for the future profession, showed creative abilities in understanding and using educational material, showed the ability to independently update and replenish knowledge; level of competence - high (creative);
«4»	is given to a student who has shown full knowledge of the curriculum, successfully completes the tasks provided by the program, mastered the basic literature recommended by the program, showed a sufficient level of knowledge in the discipline and is able to independently update and renew during further study and professional activity; level of competence - sufficient (constructive-variable);
«3»	is given to the student who has shown knowledge of the basic educational program material in the volume necessary for the further training and the subsequent work on a profession, copes with performance of the tasks provided by the program, has made separate mistakes in answers on examination and during conducting of examination tasks, but has the necessary knowledge to overcome mistakes under the guidance of a researcher; level of competence - average (reproductive);
«2»	is given to the student who did not show sufficient knowledge of the basic educational and program material, made fundamental mistakes in performance of the tasks provided by the program, cannot use the knowledge at the further training without the teacher's help, failed to master skills of independent work; level of competence - low (receptive-productive).

11. Distribution of grades obtained by applicants for higher education

The grade obtained for the differential test and the score of the average current performance during the study of the discipline are used to calculate the arithmetic mean, which is the overall grade for the discipline. The average score for the discipline is transformed into a national grade and converted into scores on a multi-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
«5»	**185-200**
«4»	**151-184**
«3»	**120-150**

Further calculations are made by the information and computer center of the University.

According to the points got on a 200-point scale, students are evaluated on a rating scale ECTS. Students enrolled in one course (one specialty) based on the number of points scored in the discipline are ranked on a scale according to ECTS system by the following way:

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

Marks ranging "A", "B", "C", "D", "E" are made by the Dean office or by any other structural department according to the Academic council decision by the educational department for the students of the definite course, which have the same specialty and **successfully** finished the discipline study.

12. Questions for the differential control on medical genetics

1. Subject and tasks of medical genetics. The role of genetics in medicine.
2. The frequency of congenital and hereditary diseases in different periods of ontogenesis. Proportion of congenital and hereditary diseases in the structure of morbidity and mortality.
3. Classification of variation. Role of modification variation in medical genetics.Genocopies. Normocopy.
4. Classification of mutations. Spontaneous and induced, somatic and germ mutations. Numerical and structural chromosomal aberrations, gene mutations. Balanced and unbalanced chromosomal aberrations.
5. Definition and classification of hereditary diseases.
6. Semiotics of hereditary diseases. Peculiarities of clinical manifestations of congenital and hereditary diseases.
7. Peculiarities of examination of the patient and his family: congenital defects, minor anomalies of the development.
8. Syndromologic approach in the diagnosis of congenital and hereditary diseases.
9. Clinical-genealogic method. Methodic of pedigree composition and analysis. Modes of inheritance.
10. Chromosomal diseases. Etiology and classification.
11. Roleof chromosomal abnormalities in ontogenesis. Pathogenesis of chromosomal diseases.
12. General symptoms of chromosomal diseases.
13. Clinical and genetic characteristics of Patau, Edwards and Down syndromes.
14. Clinical and genetic characteristics of Turner syndrome, polysomyin sex chromosomes in males and females. .
15. Clinical and genetic characteristics of syndromes with partial aneuploidy. Cri-du-chat syndrome.
16. Clinical and genetic characteristics microcytogenetic syndromes. Angelman and Prader –Willi syndromes.
17. Genomic imprinting. Definition. Genomic imprinting diseases. Etiology, pathogenesis, clinical forms. Angelman and Prader –Willi syndromes.
18. Medical-genetic counseling in chromosomal diseases. Factors of increased risk for children with chromosomal diseases.
19. Cytogenetic and molecular cytogenetic methods. Indications for cytogenetic studies.
20. Prenatal diagnosis of chromosomal diseases.
21. Monogenic (single gene) diseases. Etiology and classification.
22. Clinical polymorphism and genetic heterogeneity of monogenic disease and its causes.
23. General characteristics of autosomal-dominant diseases. The clinic, genetics and diagnosis of Marfan syndrome, Ehlers-Danlos syndrome, achondroplasia.

24. General characteristics of autosomal-recessive diseases. Factors of increased risk of having a child with an autosomal recessive disease.

25. General symptoms of inborn errors of metabolism. Principles of diagnosis. Classification of inborn errors of metabolism.

26. The clinical manifestations, genetics and diagnostics of congenital hypothyroidism, phenylketonuria, cystic fibrosis, congenital adrenal hyperplasia.

27. General characteristics of X-linked recessive diseases.

28. The clinical manifestations, genetics and diagnostics of Duchenne-Becker muscular dystrophy, fragile X syndrome.

29. Dominant X-linked disease. General characteristics. Phosphate-diabetes. Y-linked diseases.

30. Biochemical methods. Indications for biochemical diagnostics.

31. Indications for biochemical mass screening of newborns.

32. Molecular genetic methods. Indications and possibilities of this method.

33. The principles of prenatal diagnosis of single gene diseases.

34. General characteristics of mitochondrial diseases. Mitochondrial inheritance. Classification of mitochondrial diseases. General principles of diagnosis and treatment of mitochondrial diseases.

35. Diseases with hereditary predisposition (multifactorial disease). General characteristics and classification. Monogenic and polygenic forms of multifactorial diseases.

36. Hereditary based pathological reactions towards theexternal factors. Examples of pharmacogenetic and ecogenetic syndromes.

37. Oncogenetic syndromes.

38. Prevention of congenital and hereditary diseases. Types of prevention.

39. The problem of family planning and preconception prevention.

40. Medical-genetic counseling (MGC). The purpose and objectives of MGC, indications to the MGC.

41. Prenatal diagnosis (PD). Classification of the methods: invasive and noninvasive.

42. Non-invasive methods of PD. Methodic. Indications. Terms. Opportunities of the method.

43. Invasive PD. Methodic. Indications. Terms. Opportunities of the method. Contraindications. Possible complications.

44. Prenatal screening of the first and second trimester of pregnancy.

45. Screening Program. The mass (total) and selective screening programs.

46. Genetic monitoring of congenital and hereditary diseases.

Indicative list of practical work and assignments for final control 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.

13. 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 practical classes and independent work;
• Case tasks;
• Electronic data bank of MCQ tasks.

14. 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.

15. Information resources:

https://ghr.nlm.nih.gov
https://www.orpha.net
https://rarediseases.org
http://omim.org/OMIM (Online Mendelian Inheritance in Man) – An Online Catalog of Human Genes andGenetic Disorder