PRIVATE HIGHER EDUCATIONAL INSTITUTION
"INTERNATIONAL ACADEMY OF ECOLOGY AND MEDICINE"
Department of internal medicine with a course in psychiatry and narcology

WORKING PROGRAM OF EDUCATIONAL DISCIPLINE

"MEDICAL GENETICS"

LEVEL OF HIGHER EDUCATION Second (master's) level
DEGREE OF HIGHER EDUCATION Master's degree
BRANCH OF KNOWLEDGE 22 Healthcare
SPECIALTY 222 Medicine

Reviewed and approved
at the meeting of the Academic Council
Protocol No. 1, dated August 31, 2019

Kyiv 2019

Working program of education discipline "Medical Genetics" for the preparation of students of higher education of the second (master's) level of higher education in specialty 222 Medicine.

INTRODUCTION

The study program of the academic discipline "Medical Genetics" is compiled for students of the 5th year of the field of knowledge 22 "Health care" specialty 222 "Medicine"

Description of the academic discipline

<table>
<tr><td rowspan="2">Name of indicators</td><td rowspan="2">Field of knowledge, direction of training, educational qualification level</td><td>Characteristic academic discipline</td></tr>
<tr><td>Full-time teaching</td></tr>
<tr><td rowspan="2">Number of credits 1.0</td><td>Branch of knowledge 22 "Health care"</td><td rowspan="2">Full course</td></tr>
<tr><td>Specialty : 222 "Medicine"</td></tr>
<tr><td>Modules 1</td><td rowspan="4">Qualifications of the educational "Master of Medicine"</td><td>A year of training</td></tr>
<tr><td>Content modules 3</td><td>V</td></tr>
<tr><td>ECTS credits - 1.0</td><td>Semester</td></tr>
<tr><td>the total number of 30 hours</td><td>IX</td></tr>
<tr><td rowspan="8"></td><td rowspan="8">Form of education: daytime

Type of discipline: mandatory</td><td>Lectures</td></tr>
<tr><td>5 hours</td></tr>
<tr><td>Practical</td></tr>
<tr><td>15 hours</td></tr>
<tr><td>Individual work</td></tr>
<tr><td>10 hours</td></tr>
<tr><td>Type of control:</td></tr>
<tr><td>Diff. settlement</td></tr>
</table>

Note .
The ratio of the number of classroom hours to independent and individual work is (%):
for full-time study - 2: 1

PURPOSE AND OBJECTIVES OF THE DISCIPLINE

The final goals are set on the basis of the OPP training of the doctor in the specialty in accordance with the block of its content module (natural science training) and are the basis for building the content of the discipline. Description of goals is formulated through the ability in the form of goals (actions). Based on the ultimate goals of the module or the content module, specific goals are formulated in the form of specific skills (actions), objectives that ensure the achievement of the ultimate goal of studying the discipline. The end goals

are located at the beginning of the program and precede its contents, specific goals precede the content of the corresponding content module.

The ultimate goals of the discipline

- Determine the tactics of managing patients with the most common genetically determined diseases;
- Establish a preliminary diagnosis of genetically conditioned diseases associated with chromosomal aberrations and enzymopathies;
- Differentiate the most common genetically determined diseases.

As a result of studying the discipline of the student should know:

- Modern ideas about the human genome and basic concepts of genetics.
- Features of clinical manifestations of hereditary pathology, general principles of clinical diagnosis of hereditary diseases, causes of origin and diagnostic significance of morphogenetic variants.
- Phenomena of coupling and interaction of genes.
- The normal human karyotype and various types of sex determination EK4 - the structure of chromosomes and the molecular basis of mutagenesis.
- Principles of inheritance of different number of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of extranuclear genetic information.
- Genetic determinants of human blood groups and serological conflict in the Rh system.
- The value and basis of clinical-genealogical method for diagnosing decline disease, types of inheritance diseases and human traits.
- Field of application of cytogenetic methods; essence, types, possibilities of cytogenetic method in diagnostics of hereditary diseases; general characters tic chromosomal diseases, indications for use to cytogenetic sindhenan and additional special methods of examination of the patient.
- General problems of treatment, social adaptation and rehabilitation of patients with chromosomal pathology; problems of prevention of chromosomal diseases.
- Aberrations of autosomes and heterosomes that cause diseases, including oncogenesis and cancer.
- Factors affecting the primary and secondary genetic balance of the population.
- Basics of diagnostics of gene and chromosome mutations responsible for hereditary and acquired diseases, including cancer EK10 - benefits and risks of genetically modified organisms (GMOs) in the ecosystem.
- Genetic mechanisms of acquisition of drug resistance by microorganisms and cancer cells.
- Levels of biochemical diagnostics; main classes of biochemical disorders; IU Toda used for the diagnosis of metabolic; indications for biochemical genetic research.
- Principles of molecular genetic methods of diagnostics, their possibilities and limitations.
- General questions of etiology, pathogenesis, clinical genetics for monogenic hvoryuvan; indications for special methods of examination; prevention methods pocket and studied the treatment of disease pathogenesis principles first and symptomatic treatment.
- General characteristics of diseases with hereditary predisposition mechanisms D would undoubtedly hereditary predisposition, the principles of classification of the individual to the group summarized schenoho risk of a particular disease.
- Principles, stages and content of medical genetic counseling; Showing tion to sending the patient to the medical and genetic counseling.
- Principles and methods of prenatal diagnosis of hereditary and congenital diseases; indications, timing.
- Mass screening programs: appointments, conditions of holding, list of diseases to be screened.

should be able to:

- Inspect patient with hereditary diseases, identify common manifestations disease, diagnose congenital morphogenetic variants, great Vilnia use the appropriate terminology when describing clinical career slime and phenotype of the patient.
- Analyze genetic crosses and pedigrees of human traits and diseases, and assess the risk of a child being born with chromosomal aberrations.
- Identify indications for prenatal testing.
- Collect anamnestic data and genealogical information, compile a pedigree, present it graphically and analyze the type of inheritance of the disease or signs of illness in the family.
- Calculate genetic risk.
- Decide on the need for cytogenetic and molecular tests.
- Perform morphometric measurements, analyze morphogram and record karyotypes of diseases.
- Estimate the risk of manifestation of a given disease in offspring based on family predisposition and the influence of environmental factors.
- Select from the contingent of sick persons for cytogenetic, special biochemical and molecular genetic studies.
- Formulate a possible diagnosis of chromosomal pathology and some of the most common monogenic syndromes and diseases, to determine the need for additional examination, including specific genetic methods.
- Identify individuals at increased risk of multifactorial disease.
- Present the results of clinical-genetic and laboratory examination, to interpret them correctly.
- Reasonable to refer patients for medical genetic counseling, to provide the necessary documentation for medical genetic counseling.
- Carry out preventive measures aimed at preventing hereditary, congenital and multifactorial diseases, reducing the frequency of common diseases of multifactorial nature.

is ready to:

- Perceive and recognize own limitations and self-assessing educational deficits and needs.
- Use objective sources of information.
- Be guided by the well-being of a patient.
- Create and maintain close and respectful relationship with patients, as well as to demonstrate tolerance for variations in world views and cultures.

The program provides advanced knowledge of medical genetics and complements the professional knowledge of clinical residencies. It covers the issue of advances in medical genetics. Given that clinical residencies have acquired basic fundamental and clinical knowledge of the discipline and acquired some practical skills, the most recent general problems and advances in medical genetics are introduced into the lecture course. At seminars, special attention is paid to professional issues. Practical classes deepen the professional knowledge of clinical residencies in medical genetics and are dedicated to working with probands coming to the medical-genetic center. Clinical interns fill in clinical-genealogical maps, take part in ultrasound examination at prenatal diagnostics. On this basis, they make a plan for the examination of pregnant women with suspected hereditary pathology.

The formation of clinical thinking and the consolidation of practical skills by clinical interns is possible as a result of their work on specific patients.

MEDICAL GENETICS

Content module I. Heredity and pathology. Propedeutics of hereditary pathology. Prevention of hereditary pathology. Medico-genetic counseling and prenatal diagnosis

Topic 1. General characteristics of hereditary and congenital pathology. Features of manifestations of hereditary diseases.
Topic 2. Medical genetic counseling. Methods of medical genetics.
Topic 3. Prevention of hereditary and congenital pathology. Prenatal diagnosis. Screening programs.

Content module II. Chromosomal diseases. Congenital malformations.
Topic 4. General characteristics of chromosomal diseases. Clinic and diagnosis of major forms of chromosomal diseases.
Topic 5. Birth defects. Classification, etiology, diagnosis and prevention.

Content module III. Monogenic diseases.
Topic 6. General characteristics of monogenic pathology. Clinic and genetics of some forms of monogenic diseases. Diagnostic methods.
Topic 7. Systemic skeletal dysplasia (VTS).
Topic 8. Hereditary Kidney Diseases (VTS).

Content module 4. Mitochondrial diseases. Multifactorial pathology.
Topic 9. General characteristics of multifactorial diseases. Determination of genetic predisposition.
Topic 10. General characteristics of mitochondrial pathology. Clinic, diagnosis, treatment.

Types of training according to the curriculum are:

a) lectures;
b) practical classes;
c) independent work of students.

The approximate duration of the practical classes is 5 hours. The main purpose of this program is to familiarize the student with various aspects of medical genetics.
Practical classes in medical genetics include:

1. Identification of risk group for development of hereditary diseases;
2. Determination of algorithm for examination of patients with high genetic risk for development of hereditary diseases;
3. Examination of the patient for hereditary pathology, recognition of common symptoms of hereditary pathology, diagnosis of congenital morphogenetic variants, proper use of appropriate terminology in describing the clinical picture and phenotype of the patient.
4. Collection of anamnestic data and genealogical information, compilation and genealogy, presentation in graphical form, and ability to interpret the type of inheritance of the disease or signs of illness in the family.
5. Determination of the contingent of individuals who need to carry out cytogenetic, special biochemical and molecular genetic studies.
6. Ability to formulate a possible diagnosis of chromosomal pathology and some of the most common monogenic syndromes and diseases, to determine the need for additional examination, including specific genetic methods.
7. Identifying individuals at increased risk for multifactorial disease.
8. Use of clinical-genealogical method for estimation of harmful effects of environmental factors.
9. Use of methods of medical genetics for the organization of monitoring (monitoring) of the remote effects of environmental impacts.
10. Carrying out preventative measures aimed at preventing hereditary and congenital diseases.
11. Carrying out preventive measures to reduce the frequency of the most common diseases of a multifactorial nature based on genetic approaches.

Departments teaching medical genetics have the right to make changes of up to 15% to the curriculum depending on the organizational and technical capabilities, areas of research, the geo-geographical features of the region, but have to fulfill the whole range of requirements for the discipline according to the ultimate goals. OKH and OPP by specialty training and curriculum.

Students' current learning activities are monitored in practical classes in accordance with specific goals and during the individual work of the teacher with students. It is recommended to use the following means of diagnosing the level of students' preparation: computer tests, solving situational problems, conducting laboratory tests, interpreting and evaluating their results, analyzing and evaluating the results of instrumental studies and parameters that characterize the functions of the human body, its systems and organs; control of practical skills, others.

The final control of the module assimilation is carried out upon its completion.

Assessment of student achievement in the discipline is a rating and is ranked on a multi-scale scale as an assessment of mastering of the relevant module and has a definition according to the ECTS system and the traditional scale adopted in Ukraine.

CONTENTS OF THE EDUCATION PROGRAM

Module 1. Medical genetics

Content module I. Heredity and pathology. Propedeutics of hereditary pathology. Prevention of hereditary pathology. Medical - genetic counseling and prenatal diagnosis

Specific goals:

Students must :

- Know the classification of hereditary pathology.
- Explain the genetic basis of homeostasis and body variability .
- To know the frequency of congenital and hereditary pathology in different periods of ontogeny and their specific gravity in the structure of morbidity and mortality , to illustrate by example geographical and population differences in the frequencies of hereditary diseases .
- To explain the pathogenesis of inherited diseases in connection with the nature of damage to genetic structures .
- Explain the pleiotropy of genes and the multiple nature of lesions in hereditary pathology .
- Know the lethal effects of mutations (their significance in perinatal , early infant and infant mortality , infertility , spontaneous abortion).
- Explain the concepts of syndrome , association , deformity , dysplasia .
- Know the principles and stages of conducting a clinical - genealogical examination .
- Know the criteria for different types of inheritance .
- Offer schemes pedigree autosomal - dominant , autososmno - recessive , X - linked , mitochondrial type of inheritance .
- Know the morphogenetic variants and their significance in the diagnosis of hereditary syndromes and congenital conditions .
- To justify the use of cytogenetic , molecular - cytogenetic , biochemical and molecular - genetic studies.
- Know the indications and timing of prenatal diagnostics , justify the use of invasive and non-invasive methods .
- Know the indications for abortion , caused by a genetic disorder ..
- To learn the principles of organization of screening programs .
- Know the features deontoloh i chnyh and ethical issues , which may arise during the prenatal diagnosis.

The subject and tasks of medical genetics . The role of heredity in human pathology . Classification of hereditary pathology

The subject and tasks of medical genetics. The relative increase in the number of inherited diseases : population - genetic , environmental , socio - economic and demographic aspects . Classification of hereditary pathology . Ethnic , geographical , social factors , which determine the differences in the prevalence of hereditary diseases . Genetic - demographer i CHN i processes and the prevalence of hereditary diseases .

Pleiotropic action of genes and multiple lesions in hereditary pathology . Primary and secondary pleiotropes and I in the clinic for hereditary diseases . Clinical aspects pleyotrop i th , fl ' connected with syndromic and differential diagnosis nesyndromalnoyi pathology .

Semiotics and clinical diagnosis of hereditary diseases . Medico - genetic counseling (3 hrs .). Types of inheritance (autosomal - dominant , autosomal - recessive , X - and Y - linked , mitochondrial).

Semiotics of hereditary diseases . Features of manifestations of hereditary diseases . Clinical polymorphism and modifying influence of genotype on manifestations of pathological mutation . Hereditary diseases with late manifestation . Progressive character faults i gu . Affection of different organs and systems : polysystematic lesion . The concept of syndrome , association , deformity , dysplasia and her .

Medical - genetic counseling (CIM). Tasks of MGC and testimony for carrying out . Genetic risk , degree of risk . Criteria for different types of inheritance :autosomal. Principles for evaluating genetic risk in monogenic i and , chromosomal i th and multyfaktor i cial i and pathology . Organization of medical - genetic service in Ukraine . Family about both the ' object medico - genetic observation : the need to approach the family . The clinical significance of the effects of incomplete penetrance and expressivity of variation in the structure of clinical reasons, the i znoman i LTER em i deep geological common forms of hereditary diseases.

Syndromological analysis . Morphogenetic and variants of development, their genesis , postnatal modification .

Developmental Disorders: Primary and Secondary . Teratology, teratogenesis . Isolated , systemic and multiple birth defects . Consistency nature of violations stages of ontogeny.

Features of the phenotype , specificity of the spectrum of morphogenetic variants of development in hereditary and congenital pathology .

Prevention of hereditary and congenital pathology . Prenatal diagnosis . Screening programs (1 hour).

Types of prevention of hereditary diseases : primary , secondary and tertiary prevention Forms of preventive measures.

History of prenatal diagnosis . Principles of selection of nosological and diagnostics .

Content module 2. Chromosomal diseases .

Specific goals :

Students must :

- Know the etiology and cytogenetic basis of chromosomal diseases .
- Know the pathogenesis of chromosomal diseases .
- To treat cartograms in norm and at pathology .
- Know the types of disorders in the chromosomal set : structural , numeric.
- Know the characteristic clinical symptoms of chromosomal diseases .
- Know the peculiarities of clinical manifestations of individual syndromes : Down , Patau , Edward , "cat's cry" , Shereshevsky - Turner , Klinefelter .
- Learn the content , concepts , effects of chromosomal and genomic imprinting.
- To interpret the concept of single parent dyssomnia and chromosomal polymorphism.
- Explain the genetic heterogeneity of clinically similar forms of the disease.
- Know the classification , etiology , pathogenesis of birth defects .
- To master the genetic aspects of fetal growth and development , especially the embryonic and fetal

periods of pre-natal development .

- Explain the consistency of the nature of the disorders with the stages of ontogeny (gameto -, embryo , fetopathy .)

The plan of the lesson

General characteristics of chromosomal diseases . Clinic and diagnosis of major forms of chromosomal diseases. Syndrome , associated with sex chromosome abnormalities.

Human chromosomal set . The concept of karyotype .

Human chromosomal set abnormalities. Etiology and cytogenetics of chromosomal diseases . Classification , etiology , pathogenesis of chromosomal diseases . Genomic and chromosomal and gene mutations . Causes of mutations . Physical , chemical , biological mutagens . Spontaneous and induced mutagenesis (methods for studying , accounting for and controlling the mutagenic effects of anthropotogenic environmental factors). Lethal effects of mutations (their importance in perinatal i and , early childhood i and mortality , called ' links with infertility , miscarriage).

The specificity of the pathogenesis of chromosomal diseases , general patterns . Phenocytogenetic and correlations . Chromosomal aberrations and genomic mutations . Complete and mosaic forms . Parental age and frequency of chromosomal diseases in children . Lethal effects of chromosomal and genomic mutations (spontaneous abortion , stillbirth , early infant mortality).

Chromosomal diseases . Chromosomal diseases , related to quantitative changes in sex chromosomes (Turner syndrome and Klinefelter , polisomiyi). Their frequency in the population , clinical forms and variants, pathogenesis , typical clinical picture and laboratory methods for diagnosis , treatment , prognosis , rehabilitation , social adaptation .

Syndrome , associated with abnormalities of autosomes . Methods for diagnosis and treatment of chromosomal diseases , rehabilitation and social adaptation (2.5 hours) .

Chromosomal diseases , related to the quantitative changes of autosomes (Down syndrome , Patau , Edwards). Chromosomal diseases , associated with structural abnormalities of chromosomes (Syndrome Lejeune - syndrome "cry of the cat " . Microdeletion syndromes (Williams Syndrome , Prader - Willi , Anhelmana). Uniparental dysomia . Chromosomal imprytynh . Diagnosis and prevention of chromosomal diseases . Cytogenetic method . Indications conduct . The method of determining the sex chromatin . On - and metaphase chromosome analysis . Fluorescence in situ hybridization .

Content module 3. Monogenic diseases .

Specific goals :

Students must :

- To interpret the concept of monogenic diseases , hereditary metabolic diseases (CSF)
- Know the classification , general issues of the etiology and pathogenesis of monogenic diseases .
- Know the clinic , genetics , diagnosis of Ehlers - Danlo syndrome .
- Determine the leading symptom complex when evaluating the proband phenotype with Marfan syndrome .
- To determine the criteria for the diagnosis of cystic fibrosis , phenylketonuria .
- Know the clinic , genetics and diagnosis of congenital hypothyroidism .
- Know the clinic , genetics , diagnosis of hereditary metabolic disease (the disposal facility) amino acids .
- To know the clinic , genetics , diagnostics of carbohydrate chemistry .
- Know the general characteristics of bilirubin metabolism (Gilbert syndrome) .
- Know the etiology of lysosomal accumulation diseases and current diagnostic methods .
- Know the classification of hereditary kidney diseases .
- To justify the use of basic research methods in case of suspected hereditary metabolic disease (MSD

).

- Be able to interpret the results of biochemical research when clarifying the diagnosis of hereditary metabolic diseases .
- To learn the general principles of treatment of hereditary diseases , rehabilitation and social adaptation of patients .

Plan of practical class

General features of monogenic pathology. Congenital metabolic disorders (clinical picture and genetics). Amino acid metabolism disorders. Carbohydrates metabolism disorders. Disorders of bilirubin metabolism. Lysosomal storage diseases. Disorders of lipid metabolism.

General questions of monogenic diseases etiology and pathogenesis. General pathogenesis mechanisms of monogenic diseases. Pathogenesis and risk factors, asossiation with Mendelian markers. The phenomenon of anticipation.

The types of gene mutation. Genetic heterogeneity of clinically similar forms of diseases. Clinical polymorphism of etiologically single form of the disease: variable expressivity. The matter of genocopies, phenocopies and normocopies.

Classification of monogenic diseases: etiological (genetic), organ-systemic, pathogenetic.

Amino acid metabolism disorders (FKU, pathogenesis scheme). Disorders of bilirubin metabolism (Gilbert's syndrome). Carbohydrates metabolism disorders (galactosaemia, glycogenosis). Lysosomal storage diseases (mucopolysaccharidosis). Disorders of lipid metabolism (family hypercholesterolaemia). The frequency of the disorders in population, clinical forms and variants, types of mutation, pathogenesis, typical clinical picture, paraclinical and laboratory methods of diagnosing, treatment, prognosis, rehabilitation, social adaptation.

Clinical picture and genetics of several forms of monogenic diseases. The main principles of genetic diseases diagnosing and treatment, rehabilitation and social adaptation of patients (2,5 h).

Modern classification, short characteristics of the groups, the difficulties of causal classification. Pathogenesis scheme of genetic metabolic disorders.

Clinical picture and genetic of several forms of monogenic diseases with different types of inheritance. The disorders of hormone synthesis (congenital hypothyroidism, adrenogenital syndrome). Malabsorption syndrome. Celiakia. Ehlers-Danlos syndrome. Mucoviscidosis. Marphan syndrome. Fragile x syndrome. Frequency in population, clinical forms and variants, mutation types, pathogenesis, typical clinical picture, paraclinic and laboratory methods of diagnosing, treatment, prognosis, rehabilitation, social adaptation.

Opportunities of molecular genetic methods in genetic pathology diagnosing. Indications for molecular genetics observation. The value of biochemical methods in genetic metabolic disorders diagnosing. The levels of biochemical diagnostics: the primary product of gene, cellular level, metabolites in biological liquids.

Symptomatic and pathogenic therapy. The principles of pathogenic treatment as a main mean of genetic pathology treatment. Etiological treatment. Genetic engineering approach to genetic pathology treatment. Genetic therapy via somatic cells (principles, methods, results).

Context module 4. Mitochondrial diseases. Multifactor pathology.

Objectives:

Students must:

- Know the structure specialties of mitochondrial DNA and explain the principles of mitochondrial diseases development.
- Know the general characteristics of mitochondrial pathology.
- Explain the usage of different methods in mitochondrial diseases diagnosing.
- Know the general characteristics of multifactor pathologies.

- Explain susceptibility, genetic polymorphism of population.
- Illustrate monogenic determined susceptibility through examples.
- Define the criteria for polygenetic pathologies diagnosing.
- Know the genealogical, gemini, population statistical methods of multifactor pathologies analysis.
- Know the genetic principles of cancerous growth.
- Know the general characteristics of ecological factors.
- Know the side effects of pharmacological agents.
- Know the general treatment and prevention principles of influence of ecological factors and pharmacological agents.

Plan of practical class

General characteristics of mitochondrial pathology. Clinical picture, diagnostics, treatment. The features of mitochondrial DNA structure. General characteristics of mitochondrial pathology. Classification of mitochondrial diseases.

Mitochondrial inheritance. Mitochondrial diseases determined by mitochondrial DNA mutation (Laber's syndrome). Diseases determined by mitochondrial DNA deletions or duplications (clinical picture, genetics, diagnostics, therapy of Kearns-Sayre and Pearson syndromes). Diseases determined by point mutations in genes of t-RNA (clinical picture, genetics, diagnostics, therapy of MERRF, MELAS syndromes). Mitochondrial diseases determined by nuclear DNA mutations. Diseases associated with electron transport chain defects. General principles of mitochondrial pathology diagnostics snd treatment.

General characteristics of multifactor pathologies. The definition of genetic susceptibility. Preventive events.

The role of genetic and environmental factors in non-infectious pathology appearance. General characteristics of multifactor pathologies: high frequency in population; the nature of sex and age divergences; the specialties of susceptibility genes spreading and occurrence of pathology in families.

The definition of susceptibility. Genetic polymorphism of populations. Interaction between genetic susceptibility and specific conditions of the environment in context of pathology development (scheme). Exact mechanisms of genetic susceptibility realization.

Susceptibility, which is determined monogenically: ecological genetic pathology, pharmacological genetic reactions, professional diseases.

Polygenetic susceptibility as a result of non-allele genes interaction. The genetics of multifactor diseases: terminology, definition, context. Genealogical, gemini and population statistical methods in clinical and genetic analysis of multifactor diseases. The features of data collection, verification and interpretation. The relation between risk levels of multifactor pathology development and grade of relation with proband, severity of his/her condition, sex, frequency in population, professional features and mode of life. The tables of empirical risks. Susceptibility markers. The factors of increased risk.

Cancerous growth syndromes (CGS). Definition. Etiology and classification. Genetically determined forms of neoplasia. The mechanisms of CGS development. Special features of CG. The means of CGS prevention and management of the patients.

Names of modules, submodules and topics	**Number of hours**		
	Lectuers	**Practice – no classes**	**SRS**
1	2	3	4
Context module 1. General principles of medical genetics. Medical genetic counseling.			
General characteristics of congenital and genetic pathologies. Clinical features.	**1**	**2**	**-**

Medical genetic counseling. Methods in medical genetics.	-	2	-
Prevention of congenital and genetic pathologies. Prenatal diagnostics.	-	2	-
Context module 2. Chromosomal pathologies. Congenital defects.			
General characteristics of chromosomal pathologies. Clinical picture and diagnostics of main forms of chromosomal pathologies	2	1	-
Congenital defects. Classification, etiology, diagnostics, prevention	-	2	-
Context module 3. Monogenic pathologies.			
General features of monogenic pathology. Clinical picture and genetics of some monogenic pathologies. Methods of diagnosing.	1	2	-
Systemic skeletal dysplasia	-	-	1
Genetic renal disease	-	-	1
Context module 4. Mitochondrial pathology. The diseases with genetic susceptibility.			
General characteristics of mitochondrial pathology. Clinical picture, diagnostics, treatment	1	1	-
General characteristics of multifactor pathologies. Definition of genetic susceptibility	-	1	-
Individual research work	-	-	4
Preparation for practical classes	-	-	2
Final module testing control	-	2	2
TOTAL	**5**	**15**	**10**

Types of lecture

N	Topic
1.	General characteristics of congenital and genetic pathologies. Clinical features.
2.	General characteristics of chromosomal pathologies. Clinical picture and diagnostics of main forms of chromosomal pathologies
3.	General features of monogenic pathology. Clinical picture and genetics of some monogenic pathologies. Methods of diagnosing.
4.	General characteristics of mitochondrial pathology. Clinical picture, diagnostics, treatment

Types of practical classes

N	Topic
1.	General characteristics of genetic and congenital pathology. Features of genetic pathology manifestation. Medical genetic counseling. Methods in medical genetics. Prevention of genetic and congenital pathology. Prenatal diagnostics. Screening programs.
2.	General characteristics of chromosomal pathologies. Clinical picture and diagnostics of main forms of chromosomal pathologies. Congenital defects. Classification, etiology, diagnostics, prevention.
3.	General features of monogenic pathology. Clinical picture and genetics of some monogenic pathologies. Methods of diagnosing.
4.	General characteristics of mitochondrial pathology. Clinical picture, diagnostics, treatment.

	General characteristics of multifactor pathologies. Definition of genetic susceptibility
5.	Final module testing control

Types of individual work of students and its control

N	Topic	Types of control
1.	Preparation for practical classes	Current control on practical classes
2.	Individual research work	Current control on practical classes
3.	**Individual studying of topics, which are not included to auditory classes:** - systemic skeletal dysplasia - genetic renal disease	Final and current control on practical classes
4.	Preparation for final module control	Final control

THE FINAL MODULE CONTROL QUESTIONS

Context module 1. General principles of medical genetics. Medical genetic counseling.

1. The main principles of medical genetic counseling organization in Ukraine. Methodological aspects of genetic diseases diagnosing. Frequency of genetic pathology.
2. Aspects of studying medical genetics. The main diagnosing methods in medical genetics.
3. The role of genetics in pathology. Genetic diseases. Classification.
4. The semiotics of genetics diseases.
5. The features of genetic and congenital diseases clinical manifestation.
6. The main principles of genetic and congenital diseases clinical diagnosing.
7. Congenital defects.
8. Congenital morphogenetic types.
9. Syndromilogical approach in genetic and congenital diseases diagnosing.
10. The subject and objectives of medical genetics.
11. The importance of genetics in medicine.
12. The frequency of congenital and genetic pathology in different periods of ontogenesis.
13. The proportion of congenital and genetic pathology in structure of morbidity and mortality.
14. Genetic variability of congenital features as the basis of pathology.
15. The role of heredity and environment in pathology development.
16. Classification of congenital pathology.
17. The role of paraclinical research methods in congenital and genetic diagnosing.
18. Cytogenetic and molecular cytogenetic techniques. Indications for cytogenetic research.
19. Clinical genealogical method.
20. The methods of pedigree creation.
21. The main criteria of monogenic diseases inheritance (autosomal dominant, autosomal recessive, x-linked dominant and recessive). Risk group with monogenic diseases.
22. The main features of autosomal dominant inheritance type. The most wide-spread diseases.
23. The main features of autosomal recessive inheritance type. The most wide-spread diseases. Consanguine marriages.
24. The causes of genetic heterogeneity and clinic polymorphism.
25. Features of x-linked human pathology. The most wide-spread diseases.
26. The main characteristics of x-linked dominant and recessive inheritance types.
27. Y-linked type of inheritance.
28. Mitochondrial type of inheritance.
29. Biochemical methods. Indications for research.

30. Molecular genetic methods. Indications and opportunities.
31. Prevention of congenital and genetic pathology. The types of prevention.
32. The genetic base of congenital, genetic and multifactor pathology prevention.
33. The levels of prevention.
34. Family planning and preconception care.
35. Environmental care as a mean of prevention.
36. Medical genetic counseling (MGC).
37. The main objectives of MGC. Indications for MGC.
38. The functions of medical geneticists within MGC.
39. Efficiency of MGC.
40. Prenatal diagnostics (PD). The main concerns, Indications and terms of conduction.
41. Mass and selective ultrasonography screening of the pregnant.
42. Non invasive methods of PD: Indications, terms of conduction, opportunities.
43. Invasive methods of PD: indications, terms of conduction, opportunities, contraindications, possible complications.
44. Preclinical diagnostics and prevention.
45. Screening programs. Mass and selective screening programs.
46. Genetic monitoring of congenital and genetic pathology.

Content module 2. Chromosomal diseases. Congenital malformations

47. Chromosomal diseases. Etiology. Examples are common chromosomal diseases.
48. Clinical diagnosis of hereditary diseases. The concept of birth defect development, inborn morphogenetic variant.
49. Classification of birth defects (by etiology, by prevalence, depending on the stage of the provoking effect).
50. Etiology of congenital malformations.
51. The concept of gene mutation. Mutagens. Mutagenesis. Teratogenesis.
52. The concept of karyotype. Cytogenetic analysis. Indications for the study karyotype.
53. Quantitative anomalies of karyotype. Polyplodia, aneuplodia. Examples.
54. Structural anomalies of capiotype. Bapiants of structural capotypes.
55. Critical periods of human embryo development.
56. Teratogenic termination periods.
57. Genetic basis of prevention of hereditary pathology. Family Planning.
58. Chromosomal diseases. Definition of the concept. Etiology and classification.
59. Consequences of chromosomal abnormalities in ontogeny.
60. Pathogenesis of chromosomal diseases.
61. General characteristics of chromosomal diseases.
62. Genomic imprinting. Definition of the concept.
63. Clinical-genetic characteristics of Patau syndrome.
64. Clinical-genetic characteristics of Edwards syndrome.
65. Clinical-genetic characteristics of Down syndrome.
66. Clinical-genetic characteristics of trisomy 22.
67. Clinical-genetic characteristics of Shereshevsky-Turner syndrome.
68. Clinical-genetic characteristics of Kleinfelter syndrome
69. Clinical-genetic characterization of polysomes by sex chromosomes.
70. Clinical-genetic characterization of partial aneuploidy syndromes.
71. Clinical-genetic characterization of microcytogenetic syndromes.
72. Factors of increased risk of birth of children with chromosomal diseases.

Content module 3. Monogenic diseases.

73. Genetic diseases. Classification. General patterns of pathogenesis. Genocopies. Phenocopy.
74. Monogenic diseases. Definition of the concept. Etiology and classification. Risk group for monogenic diseases.
75. General patterns of pathogenesis of monogenic pathology.
76. The main features of the clinical picture of monogenic pathology.
77. Clinical polymorphism of monogenic pathology and its causes.
78. Genetic heterogeneity of monogenic diseases.
79. Clinic, genetics and diagnostics of neurofibromatosis.
80. Clinic, genetics and diagnosis of congenital hypothyroidism.
81. Clinic, genetics and diagnosis of phenylketonuria.
82. Clinic, genetics and diagnosis of cystic fibrosis.
83. Clinic, genetics and diagnosis of Marfan syndrome.
84. Clinic, genetics and diagnostics of homocystinuria.
85. Clinic, genetics and diagnostics of adrenogenital syndrome.
86. Clinics, genetics and diagnosis of Ehlers-Danloe syndrome.
87. Clinic, genetics and diagnosis of ontogenetic syndromes.
88. Diseases of genomic imprinting. Etiology, pathogenesis, clinical forms.

Content module 4. Mitochondrial pathology. Diseases with hereditary predisposition

89. General characteristics of mitochondrial pathology.
90. Classification of mitochondrial diseases.
91. Mitochondrial heredity.
92. General principles of diagnosis and treatment of mitochondrial pathology.
93. Mitochondrial diseases caused by mutations in mitochondrial DNA.
94. Clinic, genetics, diagnostics, therapy of Cairns-Sayre syndrome.
95. Clinic, genetics, diagnostics, therapy of MELAS syndrome.
96. Clinic, genetics, diagnostics, therapy of MERRF syndrome.
97. Clinic, genetics, diagnostics, therapy of Leber's syndrome.
98. Clinic, genetics, diagnostics, therapy of Pearson's syndrome.
99. Mitochondrial diseases caused by nuclear DNA mutations.
100. Diseases with hereditary predisposition. Definition of the concept. General characteristics.
101. Multifactorial pathology. Classification. General features of multi-factorial pathology. Risk group for multifactorial pathology.
102. Tendency to polygenic diseases. Inheritance factor. Twin method (concept of concordance).
103. Monogenic and polygenic forms of diseases with hereditary predisposition
104. Mechanisms of disease development with hereditary predisposition.
105. The value of hereditary predisposition in general human pathology.
106. Inherited pathological reactions to the action of external factors.
107. Ecogenetic diseases. Causes. Addiction manifestation genes from the environment.
108. Hereditary-conditioned pathological reactions to the action of external factors (atmospheric pollution, food, physical factors, metal poisoning, sensitivity to biological agents).
109. Pharmacogenetic diseases. Causes. Typical pharmaco- genetic variants.

LIST OF PRACTICAL WORKS AND TASKS FOR TOTAL MODULAR CONTROL

1. To conduct clinical-genealogical analysis of hereditary pathology. To formulate the basic tasks, indications for carrying out.

2. Demonstrate the method of genealogy assembly, graphic representation of the genealogy, using standard characters.
3. Presenting the lineage in graphical form and analyzing the type of inheritance of the disease or trait in the family
4. Demonstrate skills in collecting anamnestic data and genealogical information;
5. Demonstrate the skills of analysis and interpretation of data of genetic-genetic research methods
6. Demonstrate the skills of examination and physical examination of the patient for hereditary or congenital pathology and members of his family.
7. Formulate the principles of counseling (depending on the options of the tasks). Identify the main stages of genetic genetic counseling.
8. To conduct clinical-genealogical analysis for monogenic pathology.
9. To conduct clinical-genealogical analysis in chromosomal pathology.
10. Medico-genetic counseling for polygenic diseases.
11. Demonstrate the ability to diagnose congenital morphogenetic variants, correctly use the appropriate terminology in describing the clinical picture and phenotype of the patient;
12. Demonstrate the ability to make a preliminary diagnosis, determine the tactics of examination and management of patients with chromosomal pathology.
13. Provide a description of the patient's phenotype for chromosomal pathology or the person who has sought medical-genetic counseling
14. Be able to interpret the results of basic methods of medical genetics (cytogenetic, biochemical, molecular-genetic).
15. Provide guidance on pregnancy management and family planning based on medical history, clinical findings, and survey results.
16. To offer the scheme and algorithm of examination of patients with suspected congenital or hereditary pathology.

Control methods

Assessment is one of the final stages of learning and determining the success of learning. The grade of the discipline is given as the average of the grades for the modules for which the discipline is structured.

The module score is defined as the sum of the assessments of the current educational activity (in points) and the assessment of the final module control (in points), which is exposed when assessing theoretical knowledge and practical skills according to the lists determined by the discipline program.

The maximum number of points that a student can gain in the study of each module is 200, including 80 points (40%) for the current educational activity, and 120 points (60%) for the results of the final module control. Thus, the ratio between the results of the assessment of current learning activities and the total module control 60% to 40% is selected.

The students' current educational activities are supervised in practical classes according to specific goals. The following means of diagnostics of the level of preparation of students are recommended for application: test control (machine and without machine), solving of situational tasks, control of practical skills, in particular - the ability to properly supervise the patient, to appoint and interpret the results of laboratory and instrumental examination, to substantiate the diagnosis analysis of clinical and supportive examination methods.

In assessing the assimilation of each topic of the module, students are given grades on a 4-point (traditional) scale, using the evaluation criteria adopted by the HEI and approved by the Cycle Medical Commission. This takes into account all types of work provided by methodological development for the study of the topic.

Traditionally graded scores are converted into scores depending on the number of topics in the module so that the score for 'satisfactory' is 50-60% of the score for 'excellent'.

The maximum number of points a student can earn for his / her current activities in studying a module is calculated by multiplying the number of points corresponding to the grade "5" by the number of topics in the module by adding points for the individual student task.

The minimum number of points a student must earn when studying a module to be admitted to the final module control is calculated by multiplying the number of points corresponding to grade "3" by the number of topics in the module.

Calculation of points awarded to students in the study of the discipline "Clinical Immunology and Allergology":

I. Current control

Number of practical classes - 4

The total number of points for practical training is 80 points

(20 to 12 points per practice session)

The maximum mark for 1 day of the practical training is 20 points: 5 points for the initial level tests, 5 points for the final level tests / questions, 10 points for the practical work results during the class.

10 questions from the test database are offered to control the entry and exit levels of knowledge. Maximum student can get 5 points. The price of one question, with the right answer is 0.5 points. If a student scores less than 70% of the correct answers for the tests he / she receives 0 points.

Number of points for writing and protection of medical history - maximum 5 points (grade "5" - 5 points; grade "4" - 4 points; grade "3" - 3 points; grade "2" - 0 points).

The program used the following system to convert the traditional system into points:

Traditional System	Conversion System
"5"	20 points
"4"	16 points
"3"	12 points
"2"	0 points

The student has the right to improve their knowledge and skills in the subject, for which he received a general grade of "2" and accordingly 0 points, during the next academic session, during the current control, according to the rules of the department.

Missed material from practical classes is processed by the student on his own without scoring in accordance with this Regulation. Accordingly, the current control does not set a minimum score for admission to the final discipline control.

For class 1 pass (20% of cycle time): the lecturer is provided with a synopsis on the topic of the class missed. Passing 2 or more classes requires a second course in the discipline.

The missed subject synopsis is presented by the student to the group teacher during the next training session or after the start of study visits.

Prerequisites for issuing a synopsis of the discipline "Clinical Immunology and Allergology":

- abstract should be written by hand, not less than 10 pages of text (A4 paper), with tables and figures (diagrams)
- there must be a synopsis plan
-It should be a list of references (use literature sources since 2014, including online resources)

Final control of the course is allowed for students, provided that they attend at least 75% of classrooms (lectures, practical classes) with the total accumulated points during the study of the discipline. ***FC access to the ballpoint is not established.***

If a student has attended **less than 75% (missed more than 25% of the classes), he / she is considered to have** failed the curriculum and syllabus for the discipline, and therefore has **to re-study the discipline**.

The ability to take exams and FCs in a particular discipline is not affected by the results of other subjects.

II. Final control involves determining the level of knowledge and skills that have been formed. It is carried out upon completion of the study of discipline. Final control includes control of theoretical and practical training.

Final control (FC) is carried out at the end of the study of discipline **in the last lesson**.

The FC includes the answers to the test tasks and tasks (the execution of which controls practical skills, theoretical and practical knowledge of the discipline).

Points for final control are calculated as follows:

1. Test control: 10 theoretical questions, correct answer to 1 question - 1 point, 10 correct answers to 10 points (8% of the total FC score)
2. 10 questions, correct answer to 1 question - 1.5 points, 10 correct answers respectively 15 points (12.5% of the total FC score)
3. 10 questions in Step 2 format, correct answer to 1 question - 3.5 points, 10 correct answers to 35 points (29% of the total FC score)
4. 5 typical tasks, the correct answer to 1 task - 4 points, 5 correct answers respectively 20 points (16,50% of the total score for the FC)
5. Difficult task: The correct solution to the problem is 40 points (34% of the total FC score)

Scoring points for a task is distributed as follows:

- the diagnosis and its justification are correct, the practical tasks - 15 points (38% of the assessment for the task),
- the correct answer to the questions of diagnostics, differential diagnostics and / or pathogenesis: 13 points (32% of the total mark for solving the problem)
- Properly assigned immunotherapy (explaining the mechanisms of action of drugs) 12 points (30% of the total score for the task)

Thus, the maximum score for the FC is 120 points.

FC Score:

"5" - 97-120 points (81-100% correct answers)

"4" - 72-96 points (60-80% correct answers)

"3" - 71-61 points (59-51% correct answers)

Approved:

В.о.Ректора /Acting Rector **Dmytro GOVSIEIEV**