DANYLO HALYTSKYI LVIV NATIONAL MEDICAL UNIVERSITY

DEPARTMENT OF PROPEDEUTIC PEDIATRICS AND MEDICAL GENETICS

"APPROVED"
First Vice-Rector on Scientific and Pedagogical Work
As. Professor Iryna SOLONYNKO

"12" 06 2023

DISCIPLINE PROGRAM

OK 21.2 «MEDICAL GENETICS»

Second (master's) level of higher education
Field of Knowledge 22 "Healthcare"
Specialty 222 "Medicine"
Faculty, year: Foreign Students, 3rd year

Discussed and approved
at the educational-methodical meeting
of the Department of Propaedeutic pediatrics
and medical genetics
Minutes No 8 dated "04" 04 2023

Head of Department _____
Prof. Olena LYCHKOVSKA

Approved
by the Profile Methodical Board on
Pediatric Disciplines

Minutes No 27 dated "27" 04 2023

Head of the Board _____
Prof. Lesya BESH

The discipline program OK 21.2 «MEDICAL GENETICS» was developed and imported at the Department of Propaedeutic Pediatrics and Medical Genetics of Danylo Halytsky Lviv Nnational Medical University for the 3rd year students of Faculty Foreign Students by the Specialty 222 "Medicine".

Changes and additions to the study program of the discipline during 2023-2024 academic year.

No	Content of changes (additions)	Minutes of the meeting of the Department, date	Notes
1	No changes or additions have been made	Discussed and approved at the educational-methodical meeting of the Department of Propaedeutic Pediatrics and Medical Genetics Minutes No. 8 dated April 4, 2023	

THIS PROGRAMME WAS CREATED BY

I.Yu. Kulachkovska, associated professor of the department propaedeutic pediatrics and medical genetics Danylo Halytskyi Lviv National Medical University, Ph.D.

REVIEWER

O.B. Nadraga, Professor of Department of Pediatric Infectious Diseases, Danylo Halytskyi Lviv National Medical University, MD

Head of Department
of Propaedeutic Pediatrics
and Medical Genetics, MD, Professor Olena LYCHKOVSKA

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INTRODUCTION

The work academic programme of discipline «Medical genetics» has been set up based on the Standard of higher education in Ukraine of the second (master's) level of the field of knowledge 22 "Health care" of specialty 222 "Medicine"

The Description of the Subject (Summary)

Since heredity and variability are integral components of life, genetics should be the basis for theoretical and clinical training of the doctor. Importance of genetic knowledges for a physician is determined by a constant increasing of inherited pathology in the structure of morbidity, mortality and population disability. Studying the discipline, students should understand how to use the previously obtained theoretical knowledge from genetics in clinical practice; to enrich knowledges about medical genetics, according to the modern problems of diagnosis, treatment and prevention of hereditary pathology

The organization of the educational process is carried out according to the requirements of the European Credit Transfer System

Name of discipline	**Number of ECTS credits, number of hours, of which**				**Study year Study semester**	**Type of control**
	Totally	**Auditory**		**Self work (hr)**		
		Lectures (hr)	**Pactical classes (hr)**			
«Medical genetics» *8 Parts*	1,5 credits ECTS / 45 hr	6	16	23	III year (V / VI semesters)	Credit

The subject of discipline «Medical genetics»: clinical genetics

Interdisciplinary links: discipline «Medical genetics»:

a) It is based on the study of medical biology and genetics, medical and biological physics, medical chemistry, biological and bioorganic chemistry, morphological disciplines, normal and pathological physiology, propaedeutic of internal medicine, propaedeutic pediatrics by students and integrates with these disciplines;

b) forms the foundation for the following studying of modern diagnostic technologies that are used in the diagnosis not of hereditary diseases only, but also in clinical practice generally; trains skills to apply knowledge of modern methods of genetic diagnosis in the professional;

c)provides an understanding of the modern features of monogenic and chromosomal diseases, as well as of common human diseases that arise on the background of hereditary predisposition and require the integration of classical clinical ideas and modern high technologies.

1. The Aim and Objectives of the discipline

1.1. The Aim of the discipline «Medical genetics» is acquiring skills of active use of previously obtained theoretical knowledge in genetics in clinical practice, updating knowledge of medical and clinical genetics, especially modern problems of diagnosis, treatment and prophylaxis of hereditary pathology, studying a number of "new" common nosological forms of hereditary diseases.

1.2. **The Objectives** of studying the discipline "Medical genetics"distinguish what the student must know and be able to perform while studding the discipline.

As a result of the study of the discipline "Medical genetics" the student must know:

- the nature of hereditary human diseases, their etiology, pathogenesis, the causes of wide clinical polymorphism of etiologically uniform forms and genetic heterogeneity of clinically similar states;
- approaches and methods of detection of individuals with increased risk of multifactorial illness;
- methods of diagnostics of the most common forms of hereditary pathology;
- goals, methods and possibilities of medical genetic counseling, prenatal diagnosis and screening programs;
- goals and possibilities of modern methods of cytogenetic, biochemical and molecular genetic diagnostics;
- the principles of the interaction of the medical genetic service with all health services and indications for the organization of the flow of patients.

As a result of the study of the discipline "Medical genetics" the student must be able to:

- to examine patients and their relatives and to reveal congenital and hereditary pathology;
- to determine the clinical features of hereditary pathology and patient status;
- To estimate diagnostic, prognostic value of the manifested symptoms, and morphogenetic variants (microanomalies of development);
- to correctly collect a genetic history, to make a genealogy, to determine the type of inheritance;
- to diagnose the most common forms of hereditary pathology.

1.3 **Competency and training results,** developed by the discipline «Medical genetics» (the relationship with the normative content of the training of higher education graduates, formulated in terms of the results of study in the Standard of Higher Education).

In accordance with the requirements of the Standard of Higher Education, provides the development of the following ***competences***:

- ***integral:***

ability to solve complex problems, including those of a research and innovation nature in the field of medicine. Ability to continue studying with a high degree of autonomy.

- ***general:***

1. Ability to abstract thinking, analysis and synthesis (GC1).
2. Ability to learn and acquire modern knowledge (GC2).
3. Ability to apply knowledge in practical situations (GC3).
4. Knowledge and understanding of the subject area and understanding of professional activity (GC4).
5. Ability to adapt and act in a new situation (GC5)
6. Ability to make informed decisions (GC6)
7. Ability to work in a team (GC7).
8. The skills of interpersonal interaction (GC8).
9. Ability to use information and communication technologies (GC10)
10. Ability to search, process and analyze information from various sources (GC11)
11. Definiteness and persistence in terms of tasks and responsibilities (GC12).
12. Awareness of equal opportunities and gender problems (GC13).
13. Ability to realize one's rights and responsibilities as a member of society, to be aware of the values of a civil (free democratic) society and the nessecity for its sustainable development, the rule of law, the rights and freedoms of a person (GC14)
14. Ability to preserve and multiply moral, cultural, scientific values and achievements of society based on an understanding of the history and patterns of development of the subject area, its place in the general system of knowledge about nature and society and in the development of society, technology and technologies, to use various types and forms of motor activity for active recreation and healthy lifestyle (GC15)

- ***special (professional):***

1. Ability to gather medical information about the patient and analyze clinical data (SC1).
2. Ability to determine the required laboratory and instrumental studies and evaluate their results (SC2)
3. Ability to establish a preliminary and clinical diagnosis of the disease (SC3).
4. Ability to determine the necessary mode of study, work and rest for the treatment and prevention of diseases (SC4)
5. Ability to determine type of nourishment for the treatment and prevention of diseases (SC5)
6. Ability to determine the principles and nature of treatment of diseases (SC6)
7. Ability to assess influence of the environment, socio-economic and biological determinants on the health of an individual, family, population (SC17)
8. Convey one's own knowledge, conclusions and arguments on health care problems and related issues to specialists and non-specialists, in particular to students clearly and unequivocally (SC21)
9. Follow ethical principles during working with patients and laboratory animals (SC24)

Details of the competences are set out below in the matrix table of competences

The Matrix of Competences

No	Competence	Knowledge	Skills	Communication	Autonomy and responsibility
Integral competence					
ability to solve complex problems, including those of a research and innovation nature in the field of medicine. Ability to continue studying with a high degree of autonomy					
General competences					
1.	Ability to abstract thinking, analysis and synthesis	Know the methods of analysis, synthesis and further modern learning	Be able to analyze information, make informed decisions, be able to acquire modern knowledge.	Establish appropriate connections to achieve goals.	Be responsible for the timely acquisition of modern knowledge.
2.	Ability to learn and acquire modern knowledge	Know the current trends in the branch and analyze them	Be able to analyze professional information, make informed decisions, acquire modern knowledge.	Establish appropriate connections to achieve goals.	Be responsible for the timely acquisition of modern knowledge.
3.	Ability to apply knowledge in practical situations	Have specialized conceptual knowledge acquired in the learning process.	Be able to solve complex problems and problems that arise in professional activities	Clear and unambiguous communication of one's own conclusions, knowledge and explanations that substantiate them to specialists and non-specialists	Be responsible for making decisions in difficult conditions
4.	Knowledge and	Have deep knowledge	Be able to carry out	Ability to effectively	To be responsible for

	understanding of the subject area and understanding of professional activity	of the structure of professional activity	professional activities that require updating and integration of knowledge.	form a communication strategy in professional activities.	professional development, ability to further professional training with a high level of autonomy
5.	Ability to adapt and act in a new situation	Know the types and methods of adaptation, principles of action in a new situation.	To be able to apply means of self-regulation, to be able to adapt to new situations (circumstances) of life and activity	Establish appropriate connections to achieve results.	Be responsible for the timely use of self-regulatory methods
6.	Ability to make informed decisions	Know the tactics and strategies of communication, rules and ways of communicative behavior	Be able to make informed decisions, choose ways and strategies to communicate to ensure effective teamwork	Use communication strategies and interpersonal skills	Be responsible for the choice and tactics of communication.
7.	Ability to work in a team	Know the tactics and strategies of communication, rules and ways of communicative behavior	Be able to choose ways and strategies of communication to ensure effective teamwork	Use methods and strategies of communication to ensure effective teamwork.	Be responsible for the choice and tactics of communication.
8.	The skills of interpersonal interaction	Know the rules and methods of interpersonal interaction	Be able to choose ways and strategies of communication for interpersonal interaction	Use the skills of interpersonal interaction	Be responsible for the choice and tactics of communication.
10.	Ability to use information and communication technologies	Have deep knowledge in the field of information and communication technologies used in professional activities	Be able to use information and communication technologies in the professional field, which requires updating and integration of knowledge	Use informational and communication technologies in professional activity	Be responsible for the development of professional knowledge and skills.
11.	Ability to search, process and analyze information from various sources	Have deep knowledge in the field of information technologies used in professional activities	Be able to use information technologies in the professional field. Be able to search, and analyze information from various sources	Use informational and communication technologies in professional activity	Be responsible for the development of professional knowledge and skills.
12.	Definiteness and persistence in terms of tasks and responsibilities	Know the responsibilities and ways to accomplish the tasks.	Be able to set goals and objectives to be persistent and conscientious in the performance of duties	Establish interpersonal relationships to effectively perform tasks and responsibilities	Be responsible for the quality of the tasks.
13.	Awareness of equal opportunities and gender problems	Know social and community rights and responsibilities in context of gender problems	To form one's civic consciousness, to be able to act in accordance with it.	Ability to convey one's public and social position.	Be responsible for own civic position and activities.
14.	Ability to realize one's rights and responsibilities as a member of society, to be aware of the	Know social and community rights and responsibilities	To form one's civic consciousness, to be able to act in accordance with it.	Ability to convey one's public and social position.	Be responsible for own civic position and activities.

	values of a civil (free democratic) society and the nessecity for its sustainable development, the rule of law, the rights and freedoms of a person				
15.	Ability to preserve and multiply moral, cultural, scientific values and achievements of society based on an understanding of the history and patterns of development of the subject area, its place in the general system of knowledge about nature and society and in the development of society, technology and technologies, to use various types and forms of motor activity for active recreation and healthy lifestyle	Know the basics of ethics and deontology To know the history and patterns of development of the subject area Know the basics of a healthy lifestyle	Be able to apply ethical and deontological norms and principles in professional activities Be able to promote the basics of a healthy lifestyle in professional activities actively	The ability to convey to patients, their families, colleagues their professional position	Be responsible for the implementation of ethical and deontological norms and principles in professional activities
Special competences					
1.	Ability to gather medical information about the patient and analyze clinical data	To have specialized knowledge about child's organs and systems. Know methods and standard schemes of interviewing procedure and physical examination of the patient. Know the methods of assessing the state of intrauterine development of the fetus. Know methods of assessment of psychomotor and physical development of a child	Be able to gather data about the patient's complaints, anamnesis of illness, anamnesis of life based on algorithms and standards. Conduct physical examination of the patient. Be able to assess psychomotor and physical development of a child. Be able to assess a person's health condition (including child).	The ability to form effective communication strategy to communicate with patients and their relatives. The ability to submit information about the health condition of the child to the medical documentation	Be responsible for the quality of gathered information obtained on the basis of an interview, inspection, palpation, percussion of organs and systems and for correct assessment of the human health condition, psychomotor and physical development of a child and intrauterine development of the fetus and for the determination of appropriate measures
2.	Ability to determine the required list of laboratory and instrumental studies and evaluate their results	To have specialized knowledge about a person, his organs and systems, to know the principles of laboratory and instrumental research and evaluation of their results	Be able to analyze the results of laboratory and instrumental studies and evaluate information about the patient's diagnosis	Prescribe and evaluate the results of laboratory and instrumental research reasonably	Be responsible for the correct and timely assessment of information about results of laboratory and instrumental research in a health care institution

3.	Ability to establish a preliminary and clinical diagnosis of the disease	To have specialized knowledge about a person, his organs and systems, to know the algorithm of a diagnosis in conditions of a health care institution	Be able: • to identify leading clinical symptom or syndrome; • to establish the most probable syndrome diagnosis of the disease • to appoint laboratory and / or instrumental studies of the patient • to carry out differential diagnostics of diseases	Obtain the necessary information from a specific source and, based on its analysis, formulate relevant conclusions	Be responsible for making informed decisions and actions regarding the correctness of the established preliminary and clinical diagnosis of the disease according to ethical and legal norms
4	Ability to determine the necessary mode of study, work and rest for the treatment and prevention of diseases	To have specialized knowledge about a person, a child, his organs and systems; ethical and legal norms; algorithms and standard schemes for determining the mode of study, work and rest during treatment, based on the preliminary and clinical diagnosis of the disease (according to list 2)	Be able to determine necessary mode of study and work and rest during the treatment of the disease (according to list 2) on the basis of a preliminary and clinical diagnosis, by making a reasoned decision	Form and convey to the patient and specialists the conclusions regarding the necessary mode of study, work and rest during the treatment of the disease (according to list 2)	Be responsible for the reasonableness of prescribing a work and rest regime during the treatment of a disease (according to list 2)
5	Ability to determine type of nourishment for the treatment and prevention of diseases	To have specialized knowledge about a person, a child, his organs and systems; algorithms and standard schemes for prescribing diet for the treatment of diseases (according to list 2)	Be able to determine type of nourishment for the treatment of the disease (according to list 2) on the basis of a preliminary and clinical diagnosis	Form and convey to the patient and specialists conclusions about nourishment during the treatment of the disease (according to list 2)	Be responsible for the reasonableness of determining nourishment during the treatment of a disease (according to list 2)
6	Ability to determine the principles and nature of disease treatment	Have specialized knowledge of algorithms and standard schemes for the treatment of diseases	To be able to determine the principles and nature of disease treatment	Form and communicate to the patient and / or his parents (guardians), specialists their own conclusions about the principles and nature of treatment	Be responsible for principles and nature of disease treatment
17.	Ability to assess influence of the environment, socio-economic and biological determinants on the health of an individual, family, population	Have specialized knowledge about the negative factors of the environment and their impact on the health of a particular contingent, the relationship between the state of the environment and the health of a particular contingent; on the impact of socio-economic and biological determinants on the health of the	Identify negative environmental factors by comparing them with existing norms and standards; be able to determine the relationship between the state of the environment and the state of health of a particular contingent be able to develop preventive	Ability to assess the impact of the environment, socio-economic and biological determinants on the health of the individual, family, population	Be responsible for the timely conclusions on the state of health of the population on the basis of the negative impact of environmental factors, socio-economic and biological determinants, for the timely submission of proposals for appropriate preventive measures.

		individual, family, population. Know the principles formation of risk groups,risk area, time and risk factors	measures; be able to identify risk groups, risk areas, time of risk, risk factors; to assess the impact of socio-economic and biological determinants on the health of the individual, family, population, on the basis of epidemiological and medical-statistical studies		
21	Convey one's own knowledge, conclusions and arguments on health care problems and related issues to specialists and non-specialists, in particular to students clearly and unequivocally	To have specialized knowledge about a person, a child, diseases, their clinical manifestations, methods of prevention.	Be able to conduct a conversation about the health condition with a patient (including a child) To be able to form the patient's commitment (including child) to comply with the prescribed treatment, regime, diet.	Form a communication strategy for effective communication with the patient	To be responsible for the qualitative gathered information based on the interview and for the choice of interview tactics
24	Follow ethical principles during working with patients and laboratory animals	Know the basics of ethics and deontology	To be able to apply ethical and deontological norms and principles in professional activity	Be able to convey one's professional position to patients, their family members, and colleagues correctly	To be responsible for the implementation of ethical and deontological norms and principles in professional activity

Learning outcome

Integrative final program learning outcomes, the formation of which are facilitated by the discipline:

Compliance with standard defined learning outcomes and competencies

Learning outcome (LO)	Code of the learning outcome	Code of competence
Have thorough knowledge about the structure of professional activity. To be able to carry out professional activities that require updating and integration of knowledge. To be responsible for professional development, the ability for further professional training with a high level of autonomy	LO-1	GC1, GC2, GC3, GC4, GC5, GC6, GC7, GC8, GC10, GC11, GC12, GC13, GC14, GC15
Understanding and knowledge of fundamental and clinical biomedical sciences, at a level sufficient for solving professional tasks in the field of health care.	LO-2	GC1, GC2, GC3, GC4, GC5, GC6, GC7, GC8, GC10, GC11, GC12, GC13, GC14, GC15
Specialized conceptual knowledge, which include scientific achievements in the field of health care and are the basis for conducting research, critical understanding of problems in the field of medicine and related interdisciplinary problems	LO-3	GC1, GC2, GC3, GC4, GC5, GC6, GC7, GC8, GC10, GC11, GC12, GC13, GC14, GC15
Identify and determine 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)	LO-4	GC1, GC2, GC3, GC6, GC7, GC8; SC1, SC2, SC3, SC24
Gather complaints, anamnesis morbi and vitae, evaluate psychomotor and physical development of the patient, the state of the organs and systems of the body, based on the results of laboratory and instrumental studies,	LO-5	GC1, GC2, GC3, GC6, GC7, GC8; SC1, SC2, SC24

evaluate information about the diagnosis (according to list 4), taking into account the age of the patient		
Determine final clinical diagnosis by making a reasoned decision and analyzing received subjective and objective data of clinical, diagnostic studies, carrying out differential diagnosis, following the relevant ethical and legal norms, under the control of the chief physician in the health care institution (according to the list 2)	LO-6	GC1, GC2, GC3, GC6, GC7, GC8; SC1, SC2, SC3, SC24
Assign and analyze diagnostic studies (laboratory, functional and/or instrumental) (according to list 4) of patients with diseases of organs and body systems for differential diagnosis of diseases (according to list 2).	LO-7	GC1, GC2, GC3, GC6; SC2
Determine the nature and principles of treatment (conservative, surgical) of patients with diseases (according to list 2), taking into account the age of the patient, in the health care institution, outside its borders and at the stages of medical evacuation, including in field conditions, on the basis of a preliminary clinical diagnosis, following the relevant ethical and legal norms, by making a reasoned decision according to existing algorithms and standard schemes, if necessary - enrich the standard scheme, be able to substantiate personalized recommendations under the control of the chief physician of a medical institution	LO-9	GC1, GC2, GC3, GC6, GC7, GC8; SC5, SC6
Determine the necessary mode of study, work, rest and nutrition based on the final clinical diagnosis, following the relevant ethical and legal norms, by making a reasoned decision according to existing algorithms and standard schemes	LO-10	GC1, GC2, GC3, GC6; SC5, SC21
Assess and monitor the child's development, provide recommendations on feeding depending on age, organize preventive vaccinations according to the schedule	LO-13	GC1, GC2, GC3, GC6, GC15; SC1, SC4, SC5, SC21
Search for the necessary information in the professional literature and databases of other sources, analyze, evaluate and apply this information	LO-21	GC10, GC11
Assess the impact of the environment on the state of human health to assess the state of morbidity of the population	LO-23	GC1, GC2, GC3, GC6; SC1, SC17
Convey one's own knowledge, conclusions and arguments on health care problems and related issues to specialists and non-specialists clearly and unambiguously	LO-25	SC21

Learning outcomes for discipline

As a result of studying the discipline «Medical genetics» student **has to**:

I. Master the modern knowledge about:

- algorithm of somatogenetic examination of a patient and members of his family.
- algorithm for compilation of pedigree and legend to pedigree.
- methodology of clinical genealogical analysis of pedigree.
- methodology for work with diagnostic catalogs.
- algorithm of conducting of syndromological analysis in the process of diagnostics of hereditary pathology.
- methods of coloring chromosomes.
- types of disorders in the chromosome set: structural, numerical.
- Classification of chromosomal diseases, depending on changes, structure, number or distortion of the chromosome ploidy.
- the concept of mosaicism.
- phenomena of chromosomal aberrations.
- the concept of "chromosomal polymorphism".
- the concept of uniparental disomy.
- molecular cytogenetic research methods.
- the benefits of molecular cytogenetic studies.
- indications for cytogenetic and molecular cytogenetic studies.
- the principles of organizing screening programs.
- criteria for conducting mass and selective screening for hereditary metabolic diseases (HMD).
- basic research methods in case of suspected HMDs.
- indications for thin-layer chromatography (TLH) of amino acids and carbohydrates of blood and urine.
- indications for conducting high-quality liquid chromatography of blood and urine amino acids (Pico Tag method).
- indications for tandem mass spectrometry (MS).
- schemes and algorithms of examination of patients with suspected HMD of amino acids, carbohydrates, connective tissue, organic aciduria.
- indications for general metabolic urine screening tests.
- mechanisms of occurrence of acute metabolic disorders in the neonatal period.
- indications for the analysis of organic acids.

- indications for the study of connective tissue metabolism.
- mechanisms of occurrence of ketosis and lactic acidosis in patients with organic aciduria.
- the clinical significance of screening programs in the early diagnosis of HMD.
- programs of biochemical diagnostics of acute HMD.
- the value of routine biochemical studies in the diagnosis of HMD.
- mechanisms for the launch of metabolic decompensation in patients with HMD.
- mechanisms of occurrence of hypoglycemia in patients with organic aciduria.
- basic research methods in violation of the exchange of fatty acids.
- criteria for selecting groups of high genetic risk for the development of MHD.
- basic molecular research methods.
- indications for molecular methods of diagnostics of hereditary pathology.
- methods of direct and indirect molecular diagnostics of hereditary pathology.
- PCR method, as the basic method of molecular diagnostics.
- types of mutations according to certain molecular methods.
- Structure and functions of the nuclear and mitochondrial genome.
- modern prenatal diagnostic capabilities.
- methods of prenatal diagnosis.
- indications for invasive prenatal diagnosis.
- terms for screening of pregnant women.
- indications for the elimination of pregnancy.

. II Be able to apply knowledge in practical situations:

- To apply a systematic approach when examining a patient and his family members.
- To identify the leading clinical symptom when evaluating the proband and its family phenotype.
- To compose an algorithm for the examination of a patient with a suspicion of hereditary pathology.
- To evaluate the phenotype of proband and members of his family.
- To conduct a syndromological analysis.
- To work with diagnostic catalogs.
- To compose and analyze pedigree and legend for pedigree.
- To evaluate the nature of the inheritance of a phenotypic feature or disease according to given pedigree.
- To draw a diagram of the structure of nucleotides, intercellular and interchain bonds.
- To draw a pattern of cell cycle periods.
- To draw a scheme of mitosis.
- To draw a meiosis scheme.
- To interpret cariograms in norm and in pathology.
- To interpret the mechanisms of division of somatic and germ cells.
- To interpret methods of obtaining preparations of mitotic chromosomes.
- To draw a three-dimensional pattern. Draw a monosytic scheme.
- To draw a pattern of the appearance of structural chromosomal rearrangements.
- To compile an algorithm for conducting a molecular cytogenetic study.
- To analyze the chromatogram.
- To interpret the results of thin-layer chromatography of amino acids and carbohydrates of blood and urine.
- To nterpret the results of common metabolic urine screening tests.
- To illustrate examples of the importance of biochemical studies in refining the diagnosis of HMD.
- To compare the effectiveness of TLC, Pico Tag, GC-MS methods.
- To interpret the results of the study of connective tissue metabolism.
- To analyze Pico TAG Schedules.
- To draw a diagram of the exchange of phenylalanine.
- To draw a methionine exchange scheme.
- To analyze the results of the PCR analysis.
- To interpret the results of DNA-diagnosis of monogenic and infectious diseases.
- To analyze sonograms.
- To compile an algorithm of cytogenetic research of chorionic cells, blood lymphocytes, amniocytes.
- To analyze the results of biochemical screening.

2. Content of information of discipline «Medical genetics»

There are 1,5 ECTS credit / 45 hours for the learning of the discipline.

Program of the discipline «Medical genetics» is structured in 8 content parts.

Part 1. Heredity and pathology. The role of heredity in human pathology

Specific Goals:

−To know the frequency of congenital and hereditary pathologies in different periods of ontogenesis.

−To know the specific gravity of the congenital and hereditary pathology in the structure of morbidity and mortality.

−To master the genetic aspects of the growth and development of the fetus, especially the embryonic and fetal periods of intrauterine development.

−To know the etiology, pathogenesis, classification of birth defects.

−To explain the genetic basis of homeostasis.

−To know the classification of hereditary pathology.

−To explain the peculiarities of the pathogenesis of hereditary diseases in connection with the nature of damage to genetic structures.

−To learn content, concept, effects of chromosomal and genomic imprinting.

−To illustrate examples of clinical polymorphism and modifying effects of the genotype on the manifestation of pathological mutation.

−To know the lethal effects of mutations (their significance in perinatal, early childhood and infant mortality, association with infertility, spontaneous abortion).

−To illustrate examples of geographical and population differences in the frequency of hereditary diseases.

Topic 1. Subject and tasks of medical genetics. The role of heredity in human pathology

Subject and tasks of medical genetics. The role of medical genetic knowledge in the practical work of the doctor. The place of medical genetics in the system of medical knowledge, the relationship of medical genetics with other clinical and prophylactic disciplines. An increase in the proportion of inherited pathology in the structure of morbidity, mortality and population disability. Relative growth of the number of hereditary diseases: population-genetic, ecological, socio-economic and demographic aspects.

Classification of hereditary pathology. Mutations as etiologic factors. Genomic, chromosomal and gene mutations. Monogenic and epigenetic diseases. Ecogenetic diseases and diseases with hereditary predisposition. Chromosomal diseases. Diseases of somatic cells. Causes of mutations. Physical, chemical, biological mutagens. Spontaneous and induced mutagenesis (methods of studying, recording and controlling the mutagenic effects of anthropogenic environmental factors).

Heredity and pathogenesis. Genetic control of pathological processes. Peculiarities of the pathogenesis of hereditary diseases in connection with the nature of damage to genetic structures. Specificity of the pathogenesis of chromosomal diseases, general laws. Phenocytogenetic correlations. General mechanisms of pathogenesis of monogenic hereditary diseases. Pathogenesis of diseases with hereditary predisposition and risk factors, association with meldifying signs or markers.

Chromosomal and genomic imprinting (content, concepts, effects).

Heredity and clinical picture. Clinical polymorphism and modifying effect of the genotype on the manifestation of pathological mutation. Genetic aspects of polymorphism of hereditary pathology.

Heredity and the effects of the disease. Fatal effects of mutations (their significance in perinatal, early childhood mortality, association with infertility, spontaneous miscarriage). Hereditary pathological reactions to various drugs. Nonspecific effects of pathological mutations and chronizations of diseases. Genetic factors and recovery.

Part 2. Methods of the Medical Genetics

Specific Goals:

−To know the principles and stages of clinical and genealogical examination.

−To know the criteria for different types of inheritance.

−To suggest a pedigree chart of autosomal dominant, autosomal-recessive, X-linked, mitochondrial types of inheritance.

−To interpret the cariograms normal and with pathology.

−To know the methods of coloring of chromosomes.

−To know the types of disorders in the chromosome set: structural, numeric.

−To determine the indications for cytogenetic and molecular cytogenetic studies.

−To explain the concept of uniparenteral dysomy and chromosomal polymorphism.

−To learn the principles of organizing screening programs.

−To propose basic research methods for suspected hereditary metabolic diseases (HMDs).

− To illustrate by example the importance of biochemical studies in refining the diagnosis of HMD.

−To explain the indications for tandem mass spectrometry (MS).

−To suggest schemes and algorithm of examination of patients with suspected HMD of amino acids, carbohydrates, connective tissue, organic aciduria.

−To explain the method of PCR as the basic method of molecular diagnostics.

−To know the basic molecular research methods.

Topic 2. Clinical and genealogical methods. Cytogenetic methods. Molecular genetic methods. Biochemical methods

Stages of clinical and genealogical examination. Basic concepts: pedigree, proband, legend pedigree, simbols. Method of collecting genealogical information and its features in different types of pathology.

The value of the clinical-genealogical method in clinical practice for understanding of the nature of the disease, assessment of clinical sings, differential diagnosis of hereditary forms of pathology, studying of the genetic heterogeneity of diseases, assessment of the risk of new cases of diseases in the family, prognosis of disease and life.

Criteria for different types of inheritance: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, mitochondrial. The nature of pedigrees, the ratio of articles, the segregation of pathological signs in families. Dependence of the pedigree nature on the frequency of genes in the population. Recessive pathology and blood relativity. The concept of "sporadic case", the possible causes of "sporadic cases" in the family, mutations de novo. The phenomenon of anticipation.

Genealogical analysis in multifactorial diseases:

the dependence of the values of repeated risk on the state of the affected individual, the number of affected relatives, the degree of kinship with proband, the proportion of diseases.

Area of application of cytogenetic methods: diagnostics of hereditary pathology, study of mutation process, investigation of normal polymorphism of chromosomes.

Variants of cytogenetic research methods. The concept of a karyotype. Modern methods of chromosome research: prometaphase analysis, in situ fluorescence hybridization, autodiagnostic study, chromosomic-specific and region-specific molecular probes.

The value of the cytogenetic method in clinical practice: the diagnosis of chromosomal diseases, the diagnosis of a number of meningual diseases associated with chromosome instability, diagnosis of cancer diseases and some forms of leukemia, assessment of mutagenic effects of drugs, monitoring of the effects of damaged environmental factors.

The versatility of DNA diagnostic methods, the possibilities of their use. Characteristics of the main methodological approaches (DNA isolation, DNA restriction, blot hybridization, sequencing). PCR method (polymerase chain reaction), method of PDRF (polymorphism of lengths of restriction fragments).

Possibilities of molecular genetic methods in diagnostics of hereditary diseases. Prenatal, pre-clinical diagnosis of diseases and diagnostics of heterozygous conditions. Indications for the application of molecular genetic methods and their limitations. The latest methods of identifying mutations is the DNA chip method.

The method of PDRF for the determination of polymorphic sites.

The significance of biochemical methods in the diagnosis of hereditary metabolic diseases.

Levels of biochemical diagnostics: primary gene product, cellular level, metabolites in biological fluids.

Sifting Diagnosis: Qualitative and Quantitative Methods. List of basic methods and their brief characterization (qualitative tests with urine, paper and thin-layer chromatography of amino acids and carbohydrates in urine and blood, electrophoresis, microbiological ionizing test Gutri, fluorometry, etc.). Sifting programs of mass diagnostics of hereditary diseases and heterozygous states.

Confirmatory Diagnostics. Quantitative determination of enzymes and metabolites. Modern methods: automatic analysis of amino acids, liquid and gas chromatography, mass spectrometry, radioimmunochemical and immunoassay methods.

Indications for a biochemical study for the diagnosis of hereditary diseases.

Part 3. Propaedeutic of hereditary pathology

Specific Goals:

- *To explain the genetic heterogeneity of clinically similar forms of diseases.*
- *To illustratt examples of hereditary illness with late manifestation.*
- *To know the classification of developmental defects.*
- *To explain the corelation of character of disorders with the stages of ontogenesis (gameto-, embryo, fetopathy.)*
- *To explain the pleiotropy of the action of genes and the multiple nature of lesions in hereditary pathology.*
- *To know morphogenetic variants and values in the diagnostics of hereditary syndromes and inborn states.*
- *To explain the concept of syndrome, association, deformity, dysplasia*

.Topic 3. Semiotics of hereditary diseases. Morphogenetic developmental options. Peculiarities of manifestation of hereditary diseases. Malformations.

Semiotics of hereditary diseases. Pleiotropy of genes and multiplicity of lesions in hereditary pathology. Primary and secondary pleiotropy in the clinic of hereditary diseases. Clinical aspect of pleiotropy associated with differential diagnosis of syndromic and non-syndromic pathology.

Features of clinical examination of patients and their relatives, which contribute to the diagnosis of congenital and hereditary pathology. Features of the phenotype, specificity of the spectrum of morphogenetic variants of development in hereditary pathology. Anthropometry in the diagnosis of hereditary diseases.

Morphogenetic variants of development (microanomalies, micro-signs, signs of dizembriogenizu), their genesis, postnatal modification. General and specific morphogenetic variants: values in the diagnosis of hereditary syndromes and inborn states.

Developmental defects: primary and secondary. Isolated, systemic and multiple birth defects. Etiological heterogeneity of PDR. The concept of syndrome, association, deformation, dysplasia.

Family as an object of medical genetic observation: the need for a family approach. Clinical significance of the phenomena of incomplete penetrance and variational expressiveness in the structure of the causes of clinical variability of etiologically unified forms of hereditary pathology. Genetic heterogeneity of clinically similar forms of diseases.

Features of manifestations of hereditary diseases. Hereditary diseases with late manifestation. Progressive nature of the gap. Severity of various organs and systems: polysystem of defeat. Resistance to therapy in some forms. The consistency of

the nature of violations with the stages of ontogenesis: gameto-, embryo- and fetopathia.

Part 4. Monogenic diseases

Specific Goals:

- *To know the general issues of etiology and pathogenesis of monogenic diseases.*
- *To know the mechanism of pathogenesis of monogenic diseases.*
- *To know the classification of monogenic diseases.*
- *To know the clinic, genetics, diagnosis of Ehlers-Danlos syndrome.*
- *To determine the leading symptom complex in assessing the phenotype of proband with Marfan's syndrome.*
- *To determine the criteria for diagnosis of cystic fibrosis.*
- *To know the clinic, genetics and diagnosis of congenital hypothyroidism.*
- *To know the classification of hereditary diseases of the kidneys.*
- *To know the general characteristics of facomatosis.*
- *To know the general characteristics of oncogenetic syndromes.*

Topic 4. General characteristics of monogenic diseases. Genetics and clinical manifestation of some syndromes

Common and rare forms. Prevalence among different contingents.

General questions of aetiology and pathogenesis of monogenic diseases. Types of gene mutations. The variety of manifestations of gene mutations in clinical biochemical, molecular-genetic levels. Effects of pre and postnatal implementation of mutant genes.

Mechanisms of pathogenesis of monogenic diseases: specificity of mutations, plurality of metabolic pathways, multiplicity of functions of proteins.

Genetic heterogeneity of clinically similar forms of diseases. Aspects of heterogeneity: polialeism, sexuality (clinical examples).

Clinical polymorphism of the etiologically uniform form of the disease: variational expressiveness. Clinical diversity as a result of the interaction of the hereditary constitution and the modifying environmental factors. The concept of imprinting at the gene level.

The concept of genome, pheno- and normocopia.

Classifications of monogenic diseases: etiologic (genetic), organ-system, pathogenetic.

Monogenic syndromes of multiple birth defects. General signs. Clinical examples. Ehlers-Danlos syndromes, Marfan, Adrenogenital syndrome. Cystic fibrosis. Hypothyroidism. Hereditary kidney disease. Hereditary diseases of the skeleton. Facomatosis: general characteristics, classification. Clinic, genetics, diagnostics of neurofibromatosis, tuberous sclerosis. Prevention of neoplasia development. Tactics of patients with facomatosis. Oncogenetic syndromes (OGSs). Definition of the concept. Etiology and classification. Hereditary forms of neoplasia. Mechanism of OGS development, especially tumor growth. Ways of prevention and tactics of patient management in OGSs.

Topic 5. Hereditary renal disease

Anatomical and physiological features of kidneys and urinary system in children. Hereditary cystic kidney disease. Hereditary nephropathy. Secondary nephropathy associated with hereditary metabolic diseases. Treatment and medical genetic counseling.

Topic 6. Systemic skeletal dysplasia

Congenital and hereditary diseases of the skeleton. Classification of Systemic Skeletal Dysplasia (SSD): International and Molecular. Clinic, genetics, diagnosis of Jeune syndrome, diastrophic dysplasia, achondroplasia, imperfect osteogenesis, hypophosphatosis. Prenatal diagnosis of SSD. Treatment.

Part 5. Chromosomal diseases

Specific Goals:

- *To know the etiology and cytogenetics of chromosomal diseases.*
- *To know the pathogenesis of chromosomal diseases.*
- *To know the characteristics of chromosomal diseases.*
- *To know the modern possibilities of prenatal diagnosis.*
- *To know the indications for the elimination of pregnancy.*
- *To know the peculiarities of clinical manifestations of certain syndromes: Down, Patau, Edvard, "cat cry", Shereshevsky-Turner, Klinefelter.*

Topic 7. Overview of chromosomal diseases. Genetics and clinical manifestation of some syndromes.

Etiology and cytogenetics of chromosomal diseases. Classification of chromosomal diseases. Chromosomal aberrations and genomic mutations. Partial trisomyas and monosomes. Complete and mosaic shapes. Uniparental dysomy. Chromosomal imprinting. Age of parents and the frequency of chromosomal diseases in children.

Pathogenesis of chromosomal diseases. Dependence of the severity of the clinical picture on the chromosomal imbalance, the amount involved in the process of eu- and heterochromatin. Mechanisms of developmental disorders and developmental defects in chromosomal diseases: non-separation in meiosis, ovogenesis disturbance, decondensation of chromosomes in oocytes.

Fatal effects of chromosomal and genomic mutations (spontaneous abortion, stillbirth, early childhood mortality).

Disorders of development, involvement of different systems in the pathological process, craniofacial dysmorphia, delayed rates of psychomotor development, mental retardation, endocrinopathy. Progressive course.

Features of clinical manifestations of individual syndromes: Down, Patau, Edwards, trisomy 8, trisomy 22, cat cry, Wolf-Hirshhorn, Shereshevsky-Turner, Klinefelter, trisomy X, polysomy Y. Population frequency. Features of the course of pregnancy in chromosomal syndromes. Oncogenetic nature of chromosomal pathology. Possibilities of therapy and rehabilitation of patients. Prenatal diagnosis of chromosomal diseases.

Part 6. Hereditary metabolic diseases

Specific Goals:

—- To know the clinic, genetics, diagnostics of HMD of amino acids.
— To know the clinic, genetics, diagnostics of HMD of carbohydrates.
– To know the etiology of lysosomal storage diseases.
– To master the general principles of treatment of hereditary diseases, rehabilitation and social adaptation of patients.
- To know the general characteristics of mitochondrial pathology.
- To know the principles of classification of mitochondrial diseases.
- To suggest schemes and algorithms for examination of patients with suspected mitochondrial diseases.
- To know the general principles of diagnosis of mitochondrial diseases.
- To know the clinic, genetics, diagnosis, therapy MERRF syndrome.
- To know the clinic, genetics, diagnostics, therapy of the syndrome MELAS ..
- To know the general principles of treatment of mitochondrial diseases

Topic 8. General characteristics of hereditary metabolic diseases. Lysosomal storage diseases General characteristics of mitochondrial pathology.

Modern classification, brief description of groups, difficulties of causal classification. Scheme of pathogenesis of hereditary diseases of metabolism.

Clinics and genetics of individual forms of monogenic diseases with different types of inheritance (PKU, homocystinuria, galactosemia, glycogenoses, Gaucher disease, Niemen-Pick) Their frequency in the population, clinical forms and variants, types of mutations, pathogenesis, typical clinical picture, paraclinical and laboratory methods diagnosis, treatment, prognosis, rehabilitation, social adaptation.

Symptomatic and pathogenetic therapy. Principles of pathogenetic treatment as the main method of therapy of hereditary diseases. Etiotropic treatment.

Genetic engineering approaches to the treatment of hereditary diseases. Gene therapy through somatic cells (principles, methods, results).

General characteristics of mitochondrial pathology. Classification of mitochondrial diseases. Mitochondrial heredity Mitochondrial diseases caused by mutations of mitochondrial DNA. Diseases caused by deletions of mitochondrial DNA. Diseases caused by point mutations of mitochondrial DNA. Clinic, genetics, diagnostics, therapy of syndromes MERRF, MELAS. Pathology associated with defects in intergeneric communication, multiple syndromes of mitochondrial DNA deletions, mitochondrial DNA deletion syndrome. Mitochondrial diseases caused by mutations of nuclear DNA. Diseases associated with defects in the respiratory chain. Diseases associated with malformation of metabolism of milk and pyruvic acids, Diseases due to defects of beta-oxidation of fatty acids. Cremation cycle erosion. Fermentation of the carnitine cycle and enzymes involved in its metabolism. General principles of diagnosis and treatment of mitochondrial pathology.

Part 7. Diseases with hereditary predisposition

Specific Goals:

- To know the general characteristics of multifactorial diseases.
- To explain the concept of predisposition, genetic polymorphism of populations.
- To illustrate examples of monogenically determined predisposition.
- To determine the criteria for polygenic predisposition.
- To know the genealogy, twin and population-statistical methods of analysis of multifactorial diseases.
- To know the genetic basis of various forms of malignant growth.

Topic 9. Overview of multifactorial diseases. Determination of genetic predisposition. Measures of prevention.

The role of hereditary factors and environmental factors in the emergence of a common pathology of non-infectious etiology. General characteristics of multifactorial diseases: high frequency in the population; the nature of gender-age differences; peculiarities of propagation of gene predisposition and prevalence of diseases in families.

Concept of predisposition. Genetic polymorphism of populations. The interaction of genetic predisposition and specific environmental conditions in the development of diseases. Specific mechanisms for the implementation of hereditary predisposition.

Monogenically determined predisposition: ecogenetic pathology, pharmacogenetic reactions, occupational diseases.

Polygenic predisposition as a result of the interaction of non-pale genes. Genetics of multifactorial diseases: terminology, notion and content. Genealogical, twin and population-statistical methods in the clinical and genetic analysis of multifactorial diseases. Oral of hereditary factors and environmental factors in the emergence of a common pathology of non-infectious etiology. General characteristics of multifactorial diseases: high frequency in the population; the nature of gender-age differences; peculiarities of propagation of gene predisposition and prevalence of diseases in families.

Concept of predisposition. Genetic polymorphism of populations. The interaction of genetic predisposition and specific environmental conditions in the development of diseases. Specific mechanisms for the implementation of hereditary predisposition.

Monogenically determined predisposition: ecogenetic pathology, pharmacogenetic reactions, occupational diseases.

Polygenic predisposition as a result of the interaction of non allelic genes. Genetics of multifactorial diseases:

terminology, notion and content. Genealogical, twin and population-statistical methods in the clinical and genetic analysis of multifactorial diseases. Features of gathering, verifying and interpreting information. Dependence of risk of development of multifactorial diseases on the degree of kinship with proband, the severity of his condition, the status of proband, the frequency of the population, the nature of work and living conditions. Tables of Empirical Risk. Inclination markers. High risk factors.

Congenital malformations of multifactorial origin.

The ability to collect, verify and interpret information. Dependence of risk of development of multifactorial diseases on the degree of kinship with proband, the severity of his condition, the status of proband, the frequency of the population, the nature of work and living conditions. Tables of Empirical Risk. Inclination markers. High risk factors.

Congenital malformations of multifactorial origin.

Topic 10. Fundamentals of ecological genetics, pharmacogenetics.

The role of the environment in human evolution. Ecogenetic diseases. Etiology and pathogenesis. Classification. Nosological forms with various provoking factors (medicines, food, climate). Occupational diseases as ecogenetic in the case of small doses. Assessment of professional suitability from an ecogenetic point of view. Hereditary factors of susceptibility to infectious diseases. Hereditary pathological reactions to various drugs.

Part 8. Medical and genetic counselling, prevention of hereditary pathology. Basic principles of treatment of hereditary diseases

Specific Goals:

- *To know the levels of prophylaxis of hereditary diseases.*
- *To know the types of prophylaxis of hereditary diseases.*
- *To know the ways of carrying out preventive measures.*
- *To know the forms of preventive measures.*
- *To know the indications for conducting medical genetic counseling.*
- *To know the indications for prenatal diagnosis.*
- *To know the methods of prenatal diagnosis.*
- *To analyze the results of biochemical screening.*
- *To know the principles of selection of nosological forms subject to screening preclinical diagnostics.*
- *To know deontological questions of screening programs.*
- *To know basic principles of treatment of hereditary diseases*

Topic 11. Programs of preconception prevention and prenatal diagnostics and neonatal screening programs. Basic principles of treatment of hereditary diseases.

Ethnic, geographical, and social factors that determine the difference in the prevalence of hereditary pathology. Genetic-demographic processes and prevalence of hereditary diseases.

Types of prophylaxis of hereditary diseases: primary, secondary and tertiary prevention. Levels of prophylaxis: pregamet, prezygote, prenatal and postnatal. Ways of carrying out preventive measures: management of penetrance and expressiveness; elimination of the embryo and fetus; family planning and childbirth; environmental protection. Forms of preventive measures: medical genetic counseling; prenatal diagnosis; mass sifting programs; "genetic" medical examination of the population (registers); environmental protection and control of the mutagenicity of environmental factors.

Medical genetic counseling (MGC) as a kind of specialized medical care to the population. MGC as a medical conclusion. The objectives of the MGC and indications for the referral of patients and their families to the MGC. Prospective and retrospective counseling. Genetic risk, degree of risk. The notion of the theoretical and empirical risk. Principles of genetic risk estimation for monogeneous, chromosomal and multifactorial pathologies. Methodology of conducting MGK.

Genetic risk calculations; notification of information to patients; helping the family make a decision. Deontological and ethical issues of MGC. Interaction of doctors at MGK. Organization of medical genetic service in Ukraine. History of antenatal diagnosis. Prenatal diagnosis as a method of prevention. General indications for prenatal diagnosis.

Non-invasive methods of prenatal diagnosis. Ultrasound study: principles, indications, timing, effectiveness of diagnosis of various diseases of the fetus, assessment of the placenta, fetal sac. Determination of the level of biochemical markers (AFP, chorionic gonadotropin, etc.) in blood serum of pregnant women as screening for the detection of PVR and chromosomal diseases of the fetus.

Invasive methods. Methods of obtaining the fruit material: chorionic and placental osteoporosis, amniotic and cordocentesis. Indications, timing, contraindications and possible complications. Diagnosis of individual nosological forms.

Deontological and ethical issues that arise during prenatal diagnosis.

Sifting programs. The essence of the programs. Principles of selection of nosological forms subject to screening preclinical diagnostics. Characteristics of the basic programs of diagnosis of phenylketonuria, congenital hypothyroidism, adrenogenital syndrome. Diagnosis of heterozygous states in high genetic risk groups. Deontologic questions for promoters.

3. Structure of discipline «Medical genetics»

Topic	Lectures	Practical classes	Self work	Individual work
Part 1. Heredity and pathology. The role of heredity in human pathology				
1. Subject and tasks of medical genetics. The role of heredity in human pathology.	-	2	1	
Totally in the part 1	-	2	1	
Part 2. Methods of the medical Genetics				

2. Clinical and genealogical methods. Cytogenetic and molecular genetic methods. Biochemical methods.	-	2	2	
Totally in the part 2	-	2	2	
Part 3. Propaedeutic of hereditary pathology				
3. Semiotics of hereditary diseases. Peculiarities of manifestation of hereditary diseases. Morphogenetic developmental options. Malformations.	-	2	2	
Totally in the part 3	-	2	2	
Part 4. Monogenic diseases				
4. General characteristics of monogenic diseases. Genetics and clinical manifestation of some syndromes.	2	2	2	
5. Hereditary renal disease		-	2	
6. Systemic skeletal dysplasia		-	2	
Totally in the part 4	2	2	6	
Part 5. Chromosomal diseases				
7. Overview of chromosomal diseases. Genetics and clinical manifestation of some syndromes.	2	2	2	
Totally in the part 5	2	2	2	
Part 6. Hereditary metabolic diseases				
8. General characteristics of hereditary metabolic diseases. Lysosomal storage diseases General characteristics of mitochondrial pathology.	-	2	4	
Totally in the part 6	-	2	4	
Part 7. Diseases with hereditary predisposition				
9. Overview of multifactorial diseases. Determination of genetic predisposition. Measures of prevention.	-	2	2	
10. Fundamentals of ecological genetics, pharmacogenetics	-	-	2	
Totally in the part 7	-	2	4	
Part 8. Medical and genetic counselling, prevention of hereditary pathology. Basic principles of treatment of hereditary diseases				
11. Programs of preconception prevention and prenatal diagnostics and neonatal screening programs. Basic principles of treatment of hereditary diseases.	2	2	2	
Totally in the part 8	2	2	2	
Totally – 45 hr / 1,5 credits ECTS	6	16	23	
Final control				**Credit**
Classroom activity - 48,9%, Self work - 51,1%				

4. Lecture Curriculum of discipline «Medical genetics»

No	Topic	Hours
Part 4. Monogenic diseases		
1	General characteristics of monogenic pathology. Genetics and clinical manifestation of some monogenic diseases.	2
Part 5. Chromosomal diseases		
2.	General characteristics of chromosomal diseases. Genetics and clinical manifestation of some chromosomal diseases	2
Part 8. Medical and genetic counselling, prevention of hereditary pathology. Basic principles of treatment of hereditary diseases		
3.	Medical and genetic counselling. Prenatal diagnostics. Screening programs. Basic principles of treatment of hereditary diseases	2
	TOTAL for discipline	6

5. Practice Curriculum of discipline «Medical genetics»

No	Topic	Hours
Part 1. Heredity and pathology. The role of heredity in human pathology		
1.	Subject and tasks of medical genetics. The role of heredity in human pathology.	2
Part 2. Methods of the Medical Genetics		

2.	Clinical and genealogical methods. Cytogenetic and molecular genetic methods. Biochemical methods.	2
Part 3. Propaedeutic of hereditary diseases		
3.	Semiotics of hereditary diseases. Peculiarities of manifestation of hereditary diseases. Morphogenetic developmental options. Malformations.	2
Part 4. Monogenic diseases		
4.	General characteristics of monogenic pathology. Genetics and clinical manifestation of some syndromes.	2
Part 5. Chromosomal diseases		
5.	Overview of chromosomal diseases. Genetics and clinical manifestation of some syndromes.	2
Part 6. Hereditary metabolic diseases		
6.	General characteristics of hereditary metabolic diseases. Lysosomal storage diseases General characteristics of mitochondrial pathology.	2
Part 7. Diseases with hereditary predisposition		
7.	Overview of multifactorial diseases. Determination of genetic predisposition. Measures of prevention.	2
Part 8. Medical and genetic counselling, prevention of hereditary pathology. Basic principles of treatment of hereditary diseases		
8.	Programs of preconception prevention and prenatal diagnostics and neonatal screening programs. Basic principles of treatment of hereditary diseases.	2
TOTAL for discipline		16

6. Self-work Curriculum of discipline «Medical genetics»

No	Topic	Hours	Type of control
Part 1. Heredity and pathology. The role of heredity in human pathology			
1.	The role of heredity in human pathology	1	During classes
Part 2. Methods of the Medical Genetics			
1.	Methods of medical genetics: clinical and genealogical methods, cytogenetic and molecular genetic methods, biochemical methods.	2	During classes
Part 3. Propaedeutic of hereditary diseases			
1.1	Morphogenetic developmental options. Malformations.	2	During classes
Part 4. Monogenic diseases			
1.	Genetics and clinical manifestation of some monogenic diseases.	2	During classes
2.	Hereditary renal disease	2	
3.	Systemic skeletal dysplasia	2	
Part 5. Chromosomal diseases			
1.	Clinical manifestation of main chromosomal diseases.	2	During classes
Part 6. Hereditary metabolic diseases			
1.	Hereditary metabolic diseases: principles of treatment, rehabilitation and social adaptation	2	
2.	General characteristics of mitochondrial diseases. Clinical manifestation, diagnosis, treatment.	2	During classes
Part 7. Diseases with hereditary predisposition			
1.	Determination of genetic predisposition. Measures of prevention of multifactorial diseases.	2	During classes
2.	Fundamentals of ecological genetics, pharmacogenetics	2	
Part 8. Medical and genetic counselling, prevention of hereditary pathology. Basic principles of treatment of hereditary diseases.			
1.	Medical and genetic counselling. Prenatal diagnostics. Screening programs.	2	During classes
	TOTAL for discipline	23	

7. Individual work is not provided by the programme (according to the order of Danylo Halytskyi LNMU No. 881-з, 15.03.2022)

8. Teaching methods

In the course of teaching of the discipline “Medical genetics” such teaching methods as verbal, visual, practical, explanatory-illustrative (visual), reproductive, problem teaching, partial-research, research method are used. The method of students' individual work is also used to comprehend and master the new material with the application of knowledge in practice and the development of skills and abilities, verification and evaluation of knowledge, skills and abilities. Visual (illustrative, demonstrative) teaching methods are used, which are accessory to the verbal method, their purpose is in a brighter expression and presentation of personal opinions. Illustrative means (photo galleries, tables, models, drawings, etc.) “come to

life" with the teacher's explanation. Demonstrations are characterized by the mobility of the demonstrative means (educational film, television program or video film or its fragment, clinical observations).

Practical methods: educational, practical work in the diagnostic department of the clinical base, abstracts of students. These methods carry new educational and cognitive information and serve for the revision and formation of practical skills with the application of previously acquired knowledge.

Creative, problem research methods determine the relatively higher level of learning process. Problem-research methodology should be based on individual, creative cognitive activity of students. As it is known, the notion of "creativity" means the creation of a new, original, "scientific" product.

Problem method is related to creativity, it supposedly is on the verge between reproduction, mental formation and creativity.

Students' individual work out of control of the teacher is the independent work at home. Self-directed and individual work contributes to the development of skills of independent cognitive activity.

Creating the situation of interest in the course of teaching of the discipline "Medical genetics" is the viewing of educational video films, the use of role-plays, educational discussions, interesting clinical observations on-line. The development of students' motivation is the means of activating learning that facilitates a better knowledge deepening.

Practical classes of the discipline are carried out at the clinical base of the department of propaedeutic of pediatrics and medical genetics (Lviv Regional Children's Clinical Hospital "OHMATDYT"). Lectures are given in the conference hall of the same base. Students also have classes on the base of State Institution "Institute of Hereditary Pathology of National Academy of Medical Sciences of Ukraine". Types of educational activity of students according to the curriculum are: a) lectures, b) practical classes, c) self work of students, in the organization of which counselling teachers have a significant role. Curriculums of lectures, practical classes, and individual work provide the implementation of all topics that are part of the content of the programme in the educational process.

The topics of the lecture course reveal the problematic issues of the relevant chapters of medical genetics.

The duration of practical classes is 2 hours. Practical classes in the organization's methodology are clinical, aimed at controlling the acquisition of theoretical material and the formation of practical skills, as well as the ability to analyze and apply the obtained knowledge to solve practical problems. The main target of each practical class is the study of the basic laws of physical and psychomotor development of children of various age groups; the features of the disorders of different systems and the most common childhood diseases, as well as the principles of rational feeding and nutrition of healthy young children. Means of control are test tasks, clinical situational tasks; the control of practical skills.

The following methodology of practical classes is used:

1. Each class begins with a 10-15 minute test control conducted in order to assess the initial level of knowledge and determine the degree of students' preparation for the class.
2. Within 15-20 minutes the teacher explains and demonstrates the methodology of children's examination, introduces the students to the principles of organizing rational nutrition, etc.
3. Within 30-35 minutes the students work independently with sick children, collect anamnesis, inspect them, perform diagnostic and therapeutic manipulations, and others. During independent work, the teacher provides methodological assistance to the students and draws attention to the most important issues on the given topic of practical class.
4. Within 25-30 minutes the teacher and the students carry out a medical round of the children that the students worked with, reporting the results of their independent work. The teacher discusses and gives an explanation, emphasizes the peculiarities of one or another survey methodology, etc. During the clinical examination, the teacher controls the final level of knowledge of students.
5. While finishing a practical class, within 10-15 minutes the teacher sums up his/her conclusions, gives the students tasks for individual work, points to the basic issues of the next topic and offers a list of recommended literature for self-study.

9. Methods of control

Methods and forms of control and evaluation of students' progress in the discipline are carried out in accordance with the requirements of the program and Instructions for evaluating the students' educational activity in the context of the implementation of the European Credit Transfer System for the organization of the educational process, approved by the Ministry of Health of Ukraine (MOH Ukraine No. 08.01-47 / 10395 dated 15.04. 2014).

In assessing students' knowledge, preference is given to standardized control methods: testing (written), structured written work, work with standard medical documentation, standardized practice control exercises.

Routine check is carried out at every practical class. Preparation of the student for the class (initial stage) is checked on the basis of the answer to 10 test tasks. At the first practical lesson, these issues are included in the final control. For the correct answer for 10-9 tests the student receives 5 points, for 8-7 tests - 4 points, for 6-5 tests - 3 points, 4 and less - 0 points.

The main stage of practical training involves working in a clinic, mastering practical skills. The control of the main stage of the occupation is carried out by assessing the student's practical skills, analyzing his/her participation in the activities of the clinical department, and the ability to solve typical situational tasks. The survey is rated 12, 8, 4, 0 points.

At the final stage of the class, a summary of the practical activity of the student is made; a task is given for the student's time of individual work after the completion of the classroom part of the class. In order to assess the student's mastering of the topic, he is asked to solve three situational problems. If 3 tasks are correctly solved, the student gets 5 points, if 2 - 4 points, if 1 - 3 points. Scores obtained during the course are: scores obtained for the tests + scores received for the questions + scores obtained for the problem solving.

Recalculation of estimates from a multipoint scale is carried out as follows:

18 - 22 points - “excellent”
14 - 17 points - “good”
10 - 13 points - “satisfactory”
0 - 9 points - “unsatisfactory”

Final control of the student's acquisition of theoretical and practical material on discipline is checked on the last practical lesson according to the schedule.

Self work of the student is one of the organizational forms of study, which is regulated by the working curriculum and is performed by the student independently outside the classroom. Types of independent work of students are: preparation for practical classes, mastering practical skills of examination of a child, writing a history of illness, searching and studying additional literature and writing reports for speeches in practical classes.

10. Routine check.

Types of routine check:

- MCQ tasks
- typical situational cases
- practical skills assessment

In assessing the mastering of each topic of the current educational activity, the student is graded with a 4-point (traditional) scale, while taking into account all types of work required by the programme. A student receives an assessment on each topic. All traditional scale grades are converted into points. ***Maximal score***, which the student can get for the current educational activity on the discipline, is **200 points**.

Minimal sc ore, which the student can get for the current educational activity on the discipline, is **120 points**.

10.1. Assessment of the current educational activity

Criteria of assessing educational activity:

- 5 / “excellent” is marked in the case when the student flawlessly mastered the theoretical material of the subject, demonstrates deep and comprehensive knowledge of the topic, the main principles of scientific sources and recommended literature, logically thinks and forms an answer, freely uses the acquired theoretical knowledge in the analysis of practical material, expresses his/her attitude to certain problems, demonstrates a high level of mastering of practical skills;
- 4 / “good” is marked provided that the student has mastered the theoretical material of the class, he has the main aspects from the primary sources and the recommended literature, he reasonably teaches him; has practical skills, expresses his thoughts on certain problems, but some inaccuracies and errors are assumed in the logic of presentation of theoretical content or in the practice of practical skills;
- 3 / “satisfactory” is marked if the student has mastered the theoretical knowledge of the educational subject, is well-versed in the primary sources and recommended literature, but isn’t convincingly responsible, confuses the concept, additional questions cause the student insecurity or lack of stable knowledge; answering practical questions, reveals inaccuracies in knowledge, does not know how to evaluate facts and phenomena, associate them with future activities, make mistakes when exercising practical skills;
- 2 / “unsatisfactory” is marked in cases when the student did not master the educational material of the topic, does not know scientific facts, definitions, is almost not oriented in the primary sources and recommended literature, there is no scientific thinking, practical skills are not formed.

11. Type of final control: credit. Final control is carried out in the form of a credit based on the results of routine check and expressed in a two-point scale “passed” or “failed”. In order to pass the student must get not less than 60% of the maximal point (120 points).

12. The scheme of calculation and distribution of points that students receive:

In assessing the mastering of each topic of the current educational activity, the student is graded with a 4-point (traditional) scale, taking into account all types of work required by the program. A student receives an assessment from each topic. All traditional marks are converted into points. The calculation of the number of points is based on the student's assessment of the traditional mark during the study of the discipline during the term, by calculating the average arithmetic (CA) rounded up to two decimal places. The resulting value is converted to a multi-scale score in such way:

$$X = \frac{\mathbf{CA \ x \ 200}}{\mathbf{5}}$$

Recalculation of the average for the current educational activity on the multi-point scale of the discipline “Medical genetics”

4- point scale	200-point scale
5.00	200

4- point scale	200-point scale
4.50	180

4- point scale	200-point scale
3.99	160

4- point scale	200- point scale
3.47	139

4.97	199
4.95	198
4.92	197
4.90	196
4.87	195
4.85	194
4.82	193
4.80	192
4.77	191
4.75	190
4.72	189
4.70	188
4.67	187
4.65	186
4.62	185
4.60	184
4.57	183
4.55	182
4.52	181

4.47	179
4.45	178
4.42	177
4.40	176
4.37	175
4.35	174
4.32	173
4.30	172
4.27	171
4.24	170
4.22	169
4.19	168
4.17	167
4.14	166
4.12	165
4.09	164
4.07	163
4.04	162
4.02	161

3.97	159
3.94	158
3.92	157
3.89	156
3.87	155
3.84	154
3.82	153
3.79	152
3.77	151
3.74	150
3.72	149
3.70	148
3.67	147
3.65	146
3.62	145
3.60	144
3.57	143
3.55	142
3.52	141
3.50	140

3.45	138
3.42	137
3.40	136
3.37	135
3.35	134
3.32	133
3.30	132
3.27	131
3.25	130
3.22	129
3.20	128
3.17	127
3.15	126
3.12	125
3.10	124
3.07	123
3.05	122
3.02	121
3	120
< 3	Not enough

Self-work is evaluated during the routine check of the topic in the relevant class. Mastery of the topics that are required only in independent work is assessed at the final control.

Maximal score, which the student can get for the current educational activity on the discipline, is **200 points**.

Minimal score, which the student can get for the current educational activity on the discipline, is 1**20 points.**

Points are independently converted into both the ECTS and 4-point scale. The ECTS scores are not converted into the 4-point scale and vice versa.

Students who study in one specialty are ranked in the ECTS scale in such way:

ECTS credit	Statistic indicator
A	The best 10 % of students
B	The following 25 % of students
C	The following 30 % of students
D	The following 25 % of students
E	The last 10 % of students

A, B, C, D, E credits are ranked to the students of this course, who study in one specialty and successfully complete the study of the discipline. Students who received FX, F (“2”) scores are not included in the list of credited students. Students with an FX score after redoing automatically receive an “E” score.

Score points for students who have successfully completed the program are converted into a traditional 4-point scale by the absolute criteria listed in the table below:

Scores of the discipline	4-point scale scores
From 170 to 200 points	5
From 140 to 169 points	4
From 139 points to the minimal point (122)	3
Lower than the minimal point (122)	2

The ECTS scores is not converted into the traditional scale because the ECTS scale and the four-point scale are independent.

Objectivity of evaluation of students' educational activity is checked by statistical methods (correlation coefficient between ECTS assessment and national scale assessment).

13. Methodical support

1. Academic programme of the discipline;
2. Curriculum of the practical classes and student's self-work;
3. Methodical guidelines for the teacher;
4. Methodical guidelines of practical classes for the students;
5. Methodical guidelines for the students’ self work;
6. Test and control tasks for practical classes;
7. List of questions for final control
8. Recorded video of lectures

9. Photo-catalogs of patients with hereditary pathology
10. Photoes of karyotypes
11. • Pedigree charts

The development of test-control questions, situational tasks for interviewing and practical tasks used to diagnose the success of training is based on a list of issues that the student should learn in the course of studying the «Medical genetics» discipline. The sets of practical tasks are standardized according to the method of practical work.

14. Recommended literature

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