SYLLABUS OF DISCIPLINE «MEDICAL GENETICS»

1. GENERAL INFORMATION

Faculty	Medical No 2
Academic program (education sector, specialty, level of higher education, form of education)	Education sector 22 “Public Health” speciality 222 “Medicine” the second Master of Medicine level of high education full-time form of education
Academic year	2023- 2024
Name of discipline, code (email address on the site of Danylo Halytskyi Lviv National Medical University)	Medical genetics, OK21.2 kaf_pedpropaedeutic@meduniv.lviv.ua
Department (*name, address, phone, e-mail*)	Department of Propedeutic Pediatrics and Medical Genetics Address: 79008, Lviv, 31a, Lysenko Str Phone: +38 (032) 260-01-88 e-mail: kaf_pedpropaedeutic@meduniv.lviv.ua
Head of the department *(e-mail)*	Prof. Lychkovska Olena e-mail: Lychkovska_Olena@meduniv.lviv.ua
Year of study	The third
Semester	VI semester
Type of discipline / module (*Compulsory / optional*)	Compulsory
Teachers (*names, scientific degrees of teachers, who teach the discipline, contact e-mail*)	Sharhorodska Evgeniya, PhD, Assistant Professor Sharhorodska_Evgeniya@meduniv.lviv.ua Bakum Khrystyna, Assistant Professor Bakum_Khrystyna@meduniv.lviv.ua
Erasmus yes / no (*availability of disciplines for students in the program Erasmus +*)	no
Person responsible for Syllabus (*person who should provide comments on the Syllabus, contact e-mail*)	Kulachkovska Iryna, PhD, Associate Professor Kulachkovska_Iryna@meduniv.lviv.ua
Number of ECTS credits	1,5
Number of hours: *total* *lectures* *practical classes* *self work*	45 6 16 23
Languages of study	English
Information about consultations	During the semesters, according to the schedule, from 15.30 to 17.00 o’clock
Address, phone	Address: 79008, Lviv, 31a, Lysenko Str Phone: +38 (032) 260-01-88

2. BRIEF ANNOTATION TO THE DISCIPLINE

According to the position of modern medicine, any human pathology is to a greater or lesser extent connected with heredity. This provision is the basis of teaching and studying medical genetics as a clinical and preventive discipline. Since heredity and variability are integral components of life, genetics should be the basis of the theoretical and clinical training of a doctor. The need for genetic knowledge for a doctor is also determined by the constant increase in the specific weight of hereditary pathology in the structure of morbidity, mortality and disability of the population.

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

3. THE AIM AND OBJECTIVES OF THE DISCIPLINE

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.

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.

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 understanding of the subject area and understanding of professional activity	Have deep knowledge of the structure of professional activity	Be able to carry out professional activities that require updating and integration of knowledge.	Ability to effectively form a communication strategy in professional activities.	To be responsible for 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	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.

		behavior			
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 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	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.
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	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

	and in the development of society, technology and technologies, to use various types and forms of motor activity for active recreation and healthy lifestyle				
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 individual, family, population. Know the principles formation of risk groups,risk area, time and risk factors	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 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	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.
21	Convey one's own	To have specialized	Be able to conduct	Формувати	To be responsible for

	knowledge, conclusions and arguments on health care problems and related issues to specialists and non-specialists, in particular to students clearly and unequivocally	knowledge about a person, a child, diseases, their clinical manifestations, methods of prevention.	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.	комунікаційну стратегію для ефективного спілкування з пацієнтом	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

4. PREREQUISITES OF THE DISCIPLINE

The study of the discipline "Medical Genetics" is provided in the third year in the VI semester, when students acquire relevant knowledge in basic basic disciplines: medical biology, human anatomy and physiology, bioorganic and biological chemistry, with which the program is integrated.

Name of the subject	Acquired knowledge and skills
Biology	Knowledge of the periods of ontogenesis. Ability to make a pedigree
Anatomy	Knowledge of the proportions of body parts in children in different periods of childhood, the anatomical structure of the systems and organs of the child.
Histology	Knowledge of the stages of bookmarking, antenatal formation and histological features of the organs and systems of the child.
Biological chemistry	Knowledge of the biological role of proteins, fats, carbohydrates, vitamins, their metabolism in the body.
Physiology	Knowledge of the maturation and features of the functions of organs and systems of the child
Pathological physiology	Ability to evaluate the results of laboratory and instrumental methods of examination of the child.

5. PROGRAM LEARNING OUTCOMES

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, evaluate information about the diagnosis (according to list 4), taking into	LO-5	GC1, GC2, GC3, GC6, GC7, GC8; SC1, SC2, SC24

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

The list of studying results

Code of studying result	Content of studying result	Reference to the code of the competence matrix
Kn-1	Have specialized knowledge about the person, the child, its organs and systems, know the methods and standard schemes of interviewing and physical examination of the patient. Know the methods of assessing the fetal development. Know the stages and methods of assessment of psychomotor and physical development of the child	LO-4, LO-5, LO-6, LO-13, LO-23
Sk-1	Get data on patient complaints, history of illness and life, in a health care facility or at the location of a sick child, using the results of an interview with the child, his parents or legal representatives according to a standard scheme. Under any circumstances (in a health care facility or at the location of a sick child), using knowledge of the child's body, organs and systems, according to certain algorithms: • get information about the general condition of the child (consciousness, constitution) and appearance (examination of the skin, subcutaneous fat layer, palpation of lymph nodes, thyroid and mammary glands); • assess the psychomotor and physical development of the child; • assess cardiovascular system (examination and palpation of the heart and superficial vessels, determination of percussion borders of the heart, auscultation of the heart and blood vessels); • assess respiratory organs (examination of the chest and upper respiratory tract, palpation of the chest, percussion and auscultation of the lungs); • assess organs of abdominal cavity (examination of the abdomen, palpation and percussion of the intestines, stomach, liver, spleen, palpation of the pancreas, kidneys, pelvic organs, examination of the rectum); • assess musculoskeletal system (examination and palpation); • assess nervous system; • assess genitourinary system.	LO-4, LO-5, LO-6, LO-13, LO-23

Com-1	Effectively form a communication strategy when communicating with the patient. Note information about the child's health in the relevant medical records	LO-4, LO-5, LO-6, LO-13, LO-23
AR-1	Be responsible for the quality of received information on the basis of interviews, palpation, percussion of organs and systems and for timely assessment of human health, psychomotor and physical development of the child and fetal development and for taking appropriate measures	LO-4, LO-5, LO-6, LO-13, LO-23
Kn-2	Have specialized knowledge about man, child, its organs and systems, know the principles of laboratory and instrumental research and evaluation of their results	LO-4, LO-5, LO-6, LO-7
Sk-2	Be able to evaluate the information about the diagnosis in the health care institution, using knowledge about the child, his organs and systems, based on the results of laboratory and instrumental studies	LO-4, LO-5, LO-6, LO-7
Com-2	Obtain the necessary information and use it to make a diagnosis in a health care setting	LO-4, LO-5, LO-6, LO-7
AR-2	Be responsible for the correct and timely evaluation of information on the results of laboratory and instrumental research in a health care facility	LO-4, LO-5, LO-6, LO-7
Kn-3	Have specialized knowledge about the person, the child, his organs and systems, know the algorithm of diagnosis in a health care institution	LO-4, LO-6
Sk-3.1 Sk-3.2	In the health care institution, its subdivision: • identify and record the leading clinical symptom or syndrome by making an informed decision,using preliminary data of the patient's anamnesis, data of physical examination of the patient, knowledge of the person, his bodies and systems, adhering to the corresponding ethical and legal norms. • establish the most probable syndromic diagnosis of the disease by making an informed decision, by comparing with standards, using previous patient history and patient examination data, based on the leading clinical symptom or syndrome, using knowledge of the person, his organs and systems, according to appropriate ethical and legal norms. In a health care facility, its unit to appoint a laboratory and / or instrumental examination of the patient by making an informed decision, based on the most probable or syndromic diagnosis, according to standard schemes, using knowledge about the child, its organs and systems, adhering to relevant ethical and legal norms.	LO-4, LO-6
Com-3	On the basis of regulatory documents to keep medical records of the patient (outpatient / inpatient card, etc.).	LO-4, LO-6
AR-3	Follow ethical and legal norms, be responsible for making informed decisions and actions regarding the correctness of the established preliminary and clinical diagnosis of the disease	LO-4, LO-6
Kn-4	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)	LO-13
Sk-4	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	LO-13
Com-4	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)	LO-13
AR-4	Be responsible for the reasonableness of prescribing a work and rest regime during the treatment of a disease (according to list 2)	LO-13
Kn-5	Have specialized knowledge about the person, the child, its organs and systems; algorithms and standard schemes of nutrition in the treatment of diseases	LO-9, LO-10, LO-13
Sk-5	Be able to determine, on the basis of preliminary and clinical diagnosis, the nature of nutrition in the treatment of diseases	LO-9, LO-10, LO-13
Com-5	To form and convey to the patient, specialists conclusions about nutrition in the treatment of the disease	LO-9, LO-10, LO-13
AR-5	Be responsible for the validity of the definition of nutrition in the treatment of the disease	LO-9, LO-10, LO-13
Kn-6	Have specialized knowledge of algorithms and standard schemes for the treatment of diseases	LO-9
Sk-6	Be able to determine the principles of treatment of diseases	LO-9
Com-6	Form and convey to the patient and / or his parents (guardians), specialists their own conclusions about the principles and nature of treatment	LO-9
AR-6	Be responsible for deciding on the principles and nature of treatment	LO-9
Kn-17	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	LO-23

	environment and the health of a particular contingent; on the impact of socio-economic and biological determinants on the health of the individual, family, population. Know the principles formation of risk groups,risk area, time and risk factors	
Sk-17	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 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	LO-23
Com-17	Ability to assess the impact of the environment, socio-economic and biological determinants on the health of the individual, family, population	LO-23
AR-17	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..	LO-23
Kn-21	To have specialized knowledge about a person, a child, diseases, their clinical manifestations, methods of prevention.	LO-10, LO-13, LO-25
Sk-21	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.	LO-10, LO-13, LO-25
Com-21	Form a communication strategy for effective communication with the patient	LO-10, LO-13, LO-25
AR-21	To be responsible for the qualitative gathered information based on the interview and for the choice of interview tactics	LO-10, LO-13, LO-25
Kn-24	Know the basics of ethics and deontology	LO-4, LO-5, LO-6
Sk-24	To be able to apply ethical and deontological norms and principles in professional activity	LO-4, LO-5, LO-6
Com-24	Be able to convey one's professional position to patients, their family members, and colleagues correctly	LO-4, LO-5, LO-6
AR-24	To be responsible for the implementation of ethical and deontological norms and principles in professional activity	LO-4, LO-5, LO-6

6. FORMAT AND SCOPE OF DISCIPLINE

Study format	Full-time	
Type of training	Hours	Number of groups
Lectures	6	11
Practical classes	16	11
Seminars	-	-
Self-work	23	11

7. TOPICS AND CONTENT OF THE DISCIPLINE

Code od type of training	Topic	Studying content	Code of studying results	Teacher
L-1	General characteristics of monogenic pathology. Genetics and clinical manifestation of some monogenic diseases.	General questions of etiology and pathogenesis of monogenic diseases. Types of gene mutations. Variety of manifestations of gene mutations at clinical biochemical, molecular-genetic levels. Genetic heterogeneity of clinically similar forms of diseases. Aspects of heterogeneity: polyalelism, polylocuity (clinical examples). Clinical polymorphism of the etiologically unique form of the disease: variational expressiveness. The concept of imprinting at the genetic level. The concept of geno-, pheno- and normocopies. Classifications of monogenic diseases: etiological (genetic), organ-systemic, pathogenetic. Monogenic syndromes of multiple congenital	Kn-1, Com-1, AR-1 Kn-2, Com-2, AR-2 Kn-3, Com-3, AR-3 Kn-4, Com-4, AR-4 Kn-5, Com-5, AR-5 Kn-6, Com-6, AR-6 Kn-17, Com-17, AR-17	Sharhorodska Evgeniya, PhD, Assistant Professor

		malformations. Clinic and genetics of some forms of monogenic diseases with different types of inheritance (PKU, homocystinuria, galactosemia, glycogenosis, Gaucher disease, Neiman-Pick) Their frequency in the population, clinical forms and variants, types of mutations, pathogenesis, typical clinical picture, paraclinical and laboratory diagnostics, treatment, prognosis, rehabilitation, social adaptation. Genetically engineered approaches to the treatment of hereditary diseases. Genotherapy through somatic cells (principles, methods, results). General characteristics of mitochondrial pathology. Classification of mitochondrial diseases. Mitochondrial heredity Mitochondrial diseases caused by mutations in mitochondrial DNA. Diseases caused by deletions of mitochondrial DNA. Diseases caused by point mutations in mitochondrial DNA. Mitochondrial diseases caused by mutations in nuclear DNA. Diseases associated with defects in the respiratory chain. Diseases associated with impaired metabolism of lactic and pyruvic acids, defects in beta-oxidation of fatty acids. Enzymopathy of the Krebs cycle, the carnitine cycle and enzymes involved in its metabolism. General principles of diagnosis and treatment of mitochondrial pathology.		
L- 2	General characteristics of chromosomal diseases. Genetics and clinical manifestation of some chromosomal diseases	Etiology and cytogenetics of chromosomal diseases. Classification of chromosomal diseases Chromosomal aberrations and genomic mutations. Partial trisomies and monosomies. Complete and mosaic forms. Single parent disomies. Chromosomal imprinting. Age of parents and frequency of chromosomal diseases in children. Mechanisms of developmental disorders and developmental defects in chromosomal diseases: non-divergence in meiosis, impaired ovogenesis, decondensation of chromosomes in oocytes. Developmental defects, involvement of various systems in the pathological process, craniofacial dysmorphia, delayed psychomotor development, mental retardation, endocrinopathy. Progressive course. Oncogenetic nature of chromosomal pathology.	Kn-1, Com-1, AR-1 Kn-2, Com-2, AR-2 Kn-3, Com-3, AR-3 Kn-17, Com-17, AR-17	
L-3	Medical and genetic counselling. Prenatal diagnostics. Screening programs. Basic principles of treatment of hereditary diseases	Prenatal diagnosis of chromosomal diseases. Ethnic, geographical, social factors that cause differences in the prevalence of hereditary pathology. Genetic and demographic processes and prevalence of hereditary diseases. Types of prevention of hereditary diseases: primary, secondary and tertiary prevention. Prevention levels: pregametic, presygotic, prenatal and postnatal. Ways of preventive measures. Forms of preventive measures: medical and genetic counseling; prenatal diagnosis; mass screening programs;	Kn-1, Com-1, AR-1 Kn-2, Com-2, AR-2 Kn-3, Com-3, AR-3 Kn-4, Com-4, AR-4 Kn-5, Com-5, AR-5 Kn-6, Com-6, AR-6 Kn-17, Com-	

		"genetic" medical examination of the population (registers); environmental protection and control of mutagenicity of environmental factors. Medical and genetic counseling: tasks and indications for referring patients and their families to MGK. Prospective and retrospective counseling. Genetic risk, degree of risk. The concept of theoretical and empirical risk. Principles of genetic risk assessment in monogenic, chromosomal and multifactorial pathology. Methods of conducting IGC. Prenatal diagnosis, general indications. Non-invasive and invasive methods of prenatal diagnosis. Screening programs. The essence of programs. Possibilities of therapy of hereditary diseases and rehabilitation of patients.	17, AR-17 Kn-21, Com-21, AR-21 Kn-21, Com-21, AR-21 Kn-24, Com-24, AR-24	
P-1	Subject and tasks of medical genetics. The role of heredity in human pathology.	Specific goals: • Know the frequency of congenital and hereditary pathology in different periods of ontogenesis. • Know the share of congenital and hereditary pathology in the structure of morbidity and mortality. • Assimilate the genetic aspects of fetal growth and development, features of the embryonic and fetal periods of fetal development. • Know the etiology, pathogenesis, classification of congenital malformations. • Explain the genetic basis of homeostasis. • Know the classification of hereditary pathology. • Explain the features of the pathogenesis of hereditary diseases in connection with the nature of damage to genetic structures. • Master the content, concepts, effects of chromosomal and genomic imprinting. • Illustrate with examples clinical polymorphism and modifying the effect of genotype on the manifestations of pathological mutations. • Know the lethal effects of mutations (their significance in perinatal, early infant and child mortality, the relationship with infertility, miscarriage). • Illustrate with examples the geographical and population differences in the frequencies of hereditary diseases.	Kn-1 Sk-1 Com-1 AR-1 Kn-2 Sk-2 Com-2 AR-2 Kn-17 Sk-17 Com-17 AR-17	Sharhorodska Evgeniya, PhD, Assistant Professor Bakum Khrystyna, Assistant Professor
P-2	Clinical and genealogical methods. Cytogenetic and molecular genetic methods. Biochemical methods.	Specific goals: • Know the principles and stages of clinical and genealogical examination. • Know the criteria for different types of inheritance. • To offer pedigree schemes of autosomal dominant, autosomal recessive, X-linked, mitochondrial types of inheritance. • To interpret karyograms in norm and at pathology. • Know the methods of staining chromosomes. • Know the types of disorders in the chromosome set: structural, numerical.	Kn-1 Sk-1 Com-1 AR-1 Kn-2 Kn-2 Com-2 AR-2 Kn-3 Sk-3 Com-3 AR-3	

		• Determine indications for cytogenetic and molecular cytogenetic studies. • Interpret the concepts of single-parent disomy and chromosomal polymorphism. • Master the principles of organizing screening programs. • Master the basic research methods when hereditary metabolic diseases (HMD) are suspected. • Illustrate with examples the importance of biochemical studies in clarifying the diagnosis of HMD • Explain the indications for tandem mass spectrometry (MS). • To offer schemes and algorithm of examination of patients with suspected HMDf amino acids, carbohydrates, connective tissue, organic aciduria. • Explain the PCR method as a basic method of molecular diagnostics. • Know the basic molecular research methods.		
P-3	Semiotics of hereditary diseases. Peculiarities of manifestation of hereditary diseases. Morphogenetic developmental options. Malformations.	Specific goals: • Explain the genetic heterogeneity of clinically similar forms of disease. • Illustrate with examples of hereditary diseases with late manifestation. • Know the classification of developmental defects. • Explain the consistency of the nature of disorders with the stages of ontogenesis (gameto-, embryo-, fetopathy.) • Explain the pleiotropic action of genes and the multiple nature of the lesion in hereditary pathology. • Know the morphogenetic variants and meanings in the diagnosis of hereditary syndromes and congenital conditions. • Explain the concepts of syndrome, association, deformity, dysplasia.	Kn-1 Sk-1 Com-1 AR-1 Kn-2 Sk-2 Com-2 AR-2 Kn-3 Sk - 3 Com-3 AR-3	
P-4	General characteristics of monogenic pathology. Genetics and clinical manifestation of some syndromes.	Specific goals: • Know the general issues of etiology and pathogenesis of monogenic diseases. • Know the mechanism of pathogenesis of monogenic diseases. • Know the classification of monogenic diseases. • Know the clinic, genetics, diagnosis of Ehlers-Danlos syndrome. • Identify the leading symptom complex when assessing the phenotype of a proband with Marfan syndrome. • Define criteria for the diagnosis of cystic fibrosis. • Know the clinic, genetics and diagnosis of congenital hypothyroidism. • Know the classification of hereditary kidney disease. • Know the clinic, genetics, diagnosis of amino acid SHO. • Know the clinic, genetics, diagnosis of carbohydrate storage. • Know the general characteristics of phacomatosis. • Know the general characteristics of	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2 Kn-3 Sk-3 Com -3 AR-3	

		oncogenetic syndromes. • Know the etiology of lysosomal storage diseases. • Master the general principles of treatment of hereditary diseases, rehabilitation and social adaptation of patients.		
P-5	Overview of chromosomal diseases. Genetics and clinical manifestation of some syndromes.	Specific goals: • Know the etiology and cytogenetics of chromosomal diseases. • Know the pathogenesis of chromosomal diseases. • Know the characteristics of chromosomal diseases. • Know the modern possibilities of prenatal diagnosis. • Know the indications for the elimination of pregnancy. • Know the features of clinical manifestations of certain syndromes: Down, Patau, Edward, "cat's cry", Shereshevsky-Turner, Klinefelter.	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2 Kn-3 Sk-3 Com -3 AR-3	
P-6	General characteristics of hereditary metabolic diseases. Lysosomal storage diseases General characteristics of mitochondrial pathology.	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. • Know the general characteristics of mitochondrial pathology. • Know the principles of classification of mitochondrial diseases. • To offer schemes and algorithms of examination of patients with suspected mitochondrial diseases. • Know the general principles of diagnosis of mitochondrial diseases. • Know the clinic, genetics, diagnosis, therapy of MERRF syndrome. • Know the clinic, genetics, diagnosis, therapy of MELAS syndrome. • Know the general principles of treatment of mitochondrial diseases	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2 Kn-3 Sk-3 Com -3 AR-3 Kn-5 Sk-5 Com-5 AR-5 Kn-6 Sk-6 Com-6 AR-6	
P-7	Overview of multifactorial diseases. Determination of genetic predisposition. Measures of prevention.	Specific goals: • Know the general characteristics of multifactorial diseases. • Explain the concept of predisposition, genetic polymorphism of populations. • Illustrate with examples the monogenic tendency. • Define criteria for polygenic susceptibility. • Know the genealogical, twin and population-statistical methods of analysis of multifactorial diseases. • Know the genetic basis of various forms of malignant growth.	Kn-1,Sk-1 Com-1,AR -1 Kn-2,Sk-2 Com-2,AR-2 Kn-3,Sk-3 Com -3,AR-3 Kn-17,Sk-17 Com-17 AR-17	
P-8	Programs of preconception prevention and prenatal diagnostics and neonatal screening programs. Basic	Specific goals: • Know the levels of prevention of hereditary diseases. • Know the types of prevention of hereditary diseases.	Kn-1,Sk-1 Com-1,AR -1 Kn-2,Sk-2 Com-2,AR-2 Kn-3,Sk-3	

	principles of treatment of hereditary diseases.	• Know the ways of preventive measures. • Know the forms of preventive measures. • Know the indications for medical and genetic counseling. • Know the indications for prenatal diagnosis. • Know the methods of prenatal diagnosis. • Analyze the results of biochemical screening. • Know the principles of selection of nosological forms that are subject to screening preclinical diagnosis. • Know the deontological issues of screening programs. • Know basic principles of treatment of hereditary diseases.	Com -3,AR-3 Kn-4,Sk-4 Com-4,AR -4 Kn-5,Sk-5 Com-5,AR-5 Kn-6,Sk-6 Com-6,AR-6 Kn-17,Sk-17 Com-17, AR-17 Kn-21,Sk-21 Com-21 AR -21 Kn-24,Sk-24 Com-24 AR -24	
SW-1	The role of heredity in human pathology.	Classification of hereditary pathology. Mutations as etiological 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, accounting for and controlling the mutagenic effects of anthropotechnogenic environmental factors). Heredity and pathogenesis. Genetic control of pathological processes. Features of the pathogenesis of hereditary diseases in connection with the nature of damage to genetic structures. Specifics of pathogenesis of chromosomal diseases, general patterns. Phenocytogenetic correlations. General mechanisms of pathogenesis of monogenic hereditary diseases. Pathogenesis of diseases with hereditary predisposition and risk factors, association with mendelian traits or markers. Chromosomal and genomic imprinting (content, concepts, effects). Heredity and clinical picture. Clinical polymorphism and modifying influence of genotype on manifestations of pathological mutation. Genetic aspects of hemorrhagic pathology polymorphism. Heredity and consequences of diseases. Lethal effects of mutations (their significance in perinatal, early infant mortality, connection with infertility, miscarriage). Hereditary pathological reactions to various drugs. Nonspecific effects of pathological mutations and chronic diseases. Genetic factors and recovery.	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2 Kn-3 Sk-3 Com -3 AR-3 Kn-17 Sk-17 Com-17 AR-17	Sharhorodska Evgeniya, PhD, Assistant Professor Bakum Khrystyna, Assistant Professor
SW-2	Methods of medical genetics: clinical-genealogical, molecular-genetic, cytogenetic and biochemical methods	tages of clinical and genealogical examination. Basic concepts: pedigree, proband, pedigree legend, symbols. Methods of collecting genealogical information and its features in different types of pathology. The value of clinical and genealogical method in clinical practice to determine the nature of the disease, assessment of clinical manifestations, differential diagnosis of	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2 Kn-3	

		hereditary forms of pathology, study of genetic heterogeneity of diseases, risk assessment of new cases in the family, disease and life prognosis. Criteria of different types of inheritance: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, holandric, mitochondrial. The nature of pedigrees, sex ratios, segregation of pathological traits in families. The dependence of the nature of the pedigree on the frequency of genes in the population. Recessive pathology and blood relationship. The concept of "sporadic case", possible causes of "sporadic cases" in the family, de novo mutations. The phenomenon of anticipation. Genealogical analysis in multifactorial diseases: the dependence of the values of recurrence risk on the sex of the affected individual, the number of affected relatives, the degree of affinity with the proband, the proportion of diseases. Field of application of cytogenetic methods: diagnosis of hereditary pathology, study of mutation process, study of normal chromosome polymorphism. Variants of cytogenetic research methods. The concept of karyotype. Modern methods of chromosome research: promethase analysis, fluorescent in situ hybridization, autoradiographic research, chromosome - specific and region - specific molecular probes. The value of the cytogenetic method in clinical practice: diagnosis of chromosomal diseases, diagnosis of a number of mendelian diseases associated with chromosome instability, diagnosis of cancer and some forms of leukemia, assessment of mutagenic effects of drugs, monitoring of environmental factors. Universality of methods of DNA diagnostics, possibilities of their use. Characteristics of the main methodological approaches (DNA isolation, DNA restriction, blot hybridization, sequencing). PCR method (polymerase chain reaction), RFLP method (restriction fragment length polymorphism). Possibilities of molecular genetic methods in the diagnosis of hereditary diseases. Prenatal, preclinical diagnosis of diseases and diagnosis of heterozygous conditions. Indications for the use of molecular genetic methods and their limitations. The latest methods of mutation identification are the method of DNA chips. PDF method for determining polymorphic sites. The value of biochemical methods in the diagnosis of hereditary metabolic diseases. Levels of biochemical diagnostics: primary gene product, cellular level, metabolites in	Sk-3 Com -3 AR-3 Kn-17 Sk-17 Com-17 AR-17	

		biological fluids. Screening diagnostics: qualitative and quantitative methods. List of basic methods and their brief characteristics (qualitative tests with urine, paper and thin-layer chromatography of amino acids and carbohydrates in urine and blood, electrophoresis, microbiological inhibitory Guthrie test, fluorometry, etc.). Screening programs for mass diagnosis of hereditary diseases and heterozygous conditions. Confirmatory diagnosis. Quantitative determination of enzymes and metabolites. Modern methods: automatic amino acid analysis, liquid and gas chromatography, mass spectrometry, radioimmunochemical and enzyme-linked immunosorbent assays. Indications for biochemical research for the diagnosis of hereditary diseases.		
SW-3	Morphogenetic developmental options. Malformations.	Morphogenetic variants of development (microanomalies, micro-signs, signs of dysembryogenesis), their genesis, postnatal modification. General and specific morphogenetic variants: significance in the diagnosis of hereditary syndromes and congenital conditions. Developmental defects: primary and secondary. Isolated, systemic and multiple congenital malformations. Etiological heterogeneity of PBR. The concept of syndrome, association, deformation, dysplasia. The family as an object of medical and 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 diversity of etiologically unique 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 flow. Involvement of various organs and systems: polysystemic lesions. Resistance to therapy in some forms. Consistency of the nature of disorders with the stages of ontogenesis: gameto-, embryo- and fetopathy.	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2 Kn-3 Sk-3 Com -3 AR-3 Kn-17 Sk-17 Com-17 AR-17	
SW-4	Genetics and clinical manifestation of some monogenic diseases.	Common and rare forms. Prevalence among different contingents. General questions of etiology and pathogenesis of monogenic diseases. Types of gene mutations. Variety of manifestations of gene mutations at clinical biochemical, molecular-genetic levels. Effects of pre- and postnatal realization of mutant genes. Mechanisms of pathogenesis of monogenic diseases: specificity of mutations, multiplicity of metabolic pathways, multiplicity of protein functions. Genetic heterogeneity of clinically similar forms of diseases. Aspects of heterogeneity: polyalelism, polylocuity (clinical examples).	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2 Kn-3 Sk-3 Com -3 AR-3 Kn-17 Sk-17 Com-17	

		Clinical polymorphism of the etiologically unique form of the disease: variational expressiveness. Clinical diversity as a result of the interaction of hereditary constitution and environmental modifiers. The concept of imprinting at the genetic level. The concept of geno-, pheno- and normocopies. Classifications of monogenic diseases: etiological (genetic), organ-systemic, pathogenetic. Monogenic syndromes of multiple congenital malformations. General signs. Clinical examples. Ehlers-Danlos, Marfan syndromes, Adrenogenital syndrome. Cystic fibrosis. Hypothyroidism. Hereditary kidney disease. Hereditary skeletal diseases. Phacomatosis: general characteristics, classification. Clinic, genetics, diagnosis of neurofibromatosis, tuberous sclerosis. Prevention of neoplasia. Tactics of management of patients with phacomatosis. Oncogenetic syndromes (CSOs). Definition of the concept. Etiology and classification. Hereditary forms of neoplasia. The mechanism of CSO development, features of tumor growth. Ways of prevention and tactics of management of patients at CSOs. Epigenetic diseases.	AR-17	
SW -5	Hereditary renal disease	Features of anatomy and physiology of kidneys and urinary system in children. Hereditary cystic kidney disease. Hereditary nephropathy. Secondary nephropathy associated with inherited metabolic diseases. Treatment and medical and genetic counseling.	Kn-1,Sk-1 Com-1,AR -1 Kn-2,Sk-2 Com-2,AR-2 Kn-3,Sk-3 Com -3,AR-3 Kn-4,Sk-4 Com-4,AR -4 Kn-5,Sk-5 Com-5,AR-5 Kn-6,Sk-6 Com-6,AR-6 Kn-17,Sk-17 Com-17 AR-17	
SW -6	Systemic skeletal dysplasia	Congenital and hereditary diseases of the skeleton. Classification of systemic skeletal dysplasias (SCD): international and molecular. Clinic, genetics, diagnosis of wife's syndrome, diastrophic dysplasia, achondroplasia, imperfect osteogenesis, hypophosphatasia. Prenatal diagnosis of SCD. Treatment.	Kn-1,Sk-1 Com-1,AR -1 Kn-2,Sk-2 Com-2,AR-2 Kn-3,Sk-3 Com -3,AR-3 Kn-4,Sk-4 Com-4,AR -4 Kn-5,Sk-5 Com-5,AR-5 Kn-6,Sk-6 Com-6,AR-6 Kn-17,Sk-17 Com-17 AR-17	
SW-7	Clinical manifestations of the main chromosomal diseases.	Features of clinical manifestations of individual syndromes: Down, Patau, Edwards, trisomy 8, trisomy 22, "cat's cry", Wolf-Hirschhorn, Shereshevsky-Turner, Klinefelter, trisomy X, polysomy Y.	Kn-1,Sk-1 Com-1,AR -1 Kn-2,Sk-2 Com-2,AR-2 Kn-3,Sk-3	

		Population frequency. Features of pregnancy with chromosomal syndromes. Oncogenetic nature of chromosomal pathology. Possibilities of therapy and rehabilitation of patients. Prenatal diagnosis of chromosomal diseases.	Com -3,AR-3 Kn-4,Sk-4 Com-4,AR -4 Kn-5,Sk-5 Com-5,AR-5 Kn-6,Sk-6 Com-6,AR-6 Kn-17,Sk-17 Com-17, AR-17	
SW -8	Hereditary metabolic diseases: principles of treatment, rehabilitation and social adaptation	Modern classification, brief description of groups, difficulties of causal classification. Scheme of pathogenesis of hereditary metabolic diseases. Clinic and genetics of some forms of monogenic diseases with different types of inheritance (PKU, homocystinuria, galactosemia, glycogenosis, Gaucher disease, Neiman-Pick) Their frequency in the population, clinical forms and variants, types of mutations, pathogenesis, typical clinical picture, paraclinical and laboratory diagnostics, treatment, prognosis, rehabilitation, social adaptation. Symptomatic and pathogenetic therapy. Principles of pathogenetic treatment as the main method of treatment of hereditary diseases. Etiotropic treatment. Genetically engineered approaches to the treatment of hereditary diseases. Genotherapy through somatic cells (principles, methods, results).	Kn-1,Sk-1 Com-1,AR -1 Kn-2,Sk-2 Com-2,AR-2 Kn-3,Sk-3 Com -3,AR-3 Kn-4,Sk-4 Com-4,AR -4 Kn-5,Sk-5 Com-5,AR-5 Kn-6,Sk-6 Com-6,AR-6 Kn-17,Sk-17 Com-17 AR-17	
SW-9	General characteristics of mitochondrial diseases. Clinical manifestation, diagnosis, treatment.	General characteristics of mitochondrial pathology. Classification of mitochondrial diseases. Mitochondrial heredity Mitochondrial diseases caused by mutations in mitochondrial DNA. Diseases caused by deletions of mitochondrial DNA. Diseases caused by point mutations in mitochondrial DNA. Clinic, genetics, diagnosis, therapy of MERRF, MELAS syndromes. Pathology associated with defects in intergenomic communication - syndromes of multiple deletions of mitochondrial DNA, syndrome of deletion of mitochondrial DNA. Mitochondrial diseases caused by mutations in nuclear DNA. Diseases associated with defects of the respiratory chain .Diseases associated with impaired metabolism of lactic and pyruvic acids, Diseases caused by defects in beta-oxidation of fatty acids. Krebs cycle enzymopathy. Enzymopathy of the cycle of carnitine and enzymes involved in its metabolism. General principles of diagnosis and treatment of mitochondrial pathology.	Kn-1,Sk-1 Com-1,AR -1 Kn-2,Sk-2 Com-2,AR-2 Kn-3,Sk-3 Com -3,AR-3 Kn-4,Sk-4 Com-4,AR -4 Kn-5,Sk-5 Com-5,AR-5 Kn-6,Sk-6 Com-6,AR-6 Kn-17,Sk-17 Com-17 AR-17	
SW-10	Determination of genetic predisposition. Measures of prevention of multifactorial diseases.	The role of hereditary and environmental factors in the occurrence of common pathology of non-infectious etiology. General characteristics of multifactorial diseases: high frequency in the population; the nature of sex and age differences; features of the spread of predisposition genes and the prevalence of diseases in families.	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2	

		The concept of predisposition. Genetic polymorphism of populations. Interaction of genetic predisposition and specific environmental conditions in the development of diseases. Specific mechanisms for the implementation of hereditary predisposition. Monogenic predisposition: ecogenetic pathology, pharmacogenetic reactions, occupational diseases. Polygenic predisposition as a result of the interaction of non-allelic genes. Genetics of multifactorial diseases: terminology, concept and content. Genealogical, twin and population-statistical methods in clinical and genetic analysis of multifactorial diseases. Features of collection, verification and interpretation of information. Dependence of the degree of risk of development of multifactorial diseases on the degree of affinity with the proband, severity of his condition, sex of the proband, population frequency, nature of work and living conditions. Empirical risk tables. Predisposition markers. High risk factors. Congenital malformations of multifactorial origin.	Kn-3 Sk-3 Com -3 AR-3 Kn-4 Sk-4 Com-4 AR -4 Kn-5 Sk-5 Com-5 AR-5 Kn-6 Sk-6 Com-6 AR-6 Kn-17 Sk-17 Com-17 AR-17	
SW-11	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.	Kn-1,Sk-1 Com-1,AR -1 Kn-2,Sk-2 Com-2,AR-2 Kn-3,Sk-3 Com -3,AR-3 Kn-4,Sk-4 Com-4,AR -4 Kn-5,Sk-5 Com-5,AR-5 Kn-6,Sk-6 Com-6,AR-6 Kn-17,Sk-17 Com-17 AR-17	
SW-12	Medical and genetic counseling. Prenatal diagnosis. Screening programs	Medical and genetic counseling (MGC) as a type of specialized medical care for the population. IGC as a medical opinion. Tasks of IGC and indications for referral of patients and their families to IGC. Prospective and retrospective counseling. Genetic risk, degree of risk. The concept of theoretical and empirical risk. Principles of genetic risk assessment in monogenic, chromosomal and multifactorial pathology. Methods of conducting IGC. Genetic risk calculations; communication of information to patients; assisting the family in making decisions. Deontological and ethical issues of IGC. Interaction of doctors at MGK. Organization of medical and genetic service in Ukraine. History of prenatal diagnostics. Prenatal diagnosis as a method of prevention. General indications for prenatal diagnosis. Non-invasive methods of prenatal diagnosis. Ultrasound examination: principles, indications, timing, effectiveness of diagnosis of various diseases of the fetus, assessment of	Kn-1 Sk-1 Com-1 AR -1 Kn-2 Sk-2 Com-2 AR-2 Kn-3 Sk-3 Com -3 AR-3 Kn-4 Sk-4 Com-4 AR -4 Kn-5 Sk-5 Com-5 AR-5 Kn-6 Sk-6 Com-6	

		the condition of the placenta, amniotic sac. Determination of the level of biochemical markers (AFP, chorionic gonadotropin, etc.) in the serum of pregnant women as a screening for the detection of PVR and chromosomal diseases of the fetus. Invasive methods. Methods of obtaining fetal material: chorionic and placentobiopsy, amniocentesis and cordocentesis. Indications, terms, contraindications and possible complications. Diagnosis of certain nosological forms. Deontological and ethical issues that arise during prenatal diagnosis. Screening programs. The essence of programs. Principles of selection of nosological forms subject to screening preclinical diagnosis. Characteristics of the main programs for the diagnosis of phenylketonuria, congenital hypothyroidism, adrenogenital syndrome. Diagnosis of heterozygous conditions in high genetic risk groups. Deontological issues of screening programs	AR-6 Kn-17 Sk-17 Com-17 AR-17 Kn-21 Sk-21 Com-21 AR -21 Kn-24 Sk-24 Com-24 AR -24	

Teaching methods and system of organization of classes

Teaching the discipline "Medical genetics" consists of verbal, visual, practical, explanatory-illustrative (visual), reproductive, problem teaching, part-search, research methods. Method of independent work of students is used to understand and master the new material of work on 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 auxiliary to the verbal method, their significance is to provide a brighter presentation and self-reflection. Illustrations (photo galleries, tables, models, images, etc.) are used also. Following types of demonstration: educational film, television program or video are very helpful for education.

Practical methods: educational, practical work in the diagnostic department of the hospital, student abstracts. These methods carry new educational and cognitive information and serve for consolidation, the formation of practical skills in the application of previously acquired knowledge.

Creative, problem-searching methods determine the relatively higher level of studing process. Problem-search methodology should be based on independent, creative cognitive activity of students. The concept of "creativity" is the creation of a new, original, "scientific" product.

Problem method of teaching is close to creativity, it is situated between reproduction, mental formation and creativity.

Independent work of students outside the control of a teacher - independent work at home. Self-directed and individual work contributes to the development of skills for independent cognitive activity.

Creating a situation of interest in the teaching of discipline "Medical genetics" - a review of educational video, using role-playing games, educational discussions, interesting clinical observations in the on-line system). The development of students' motivation means activating learning that facilitates a better studying.

Types of educational activity of students according to the curriculum are: a) lectures, b) practical classes, c) self-work of students.. Thematic plans of lectures, practical classes, self-work provide the implementation of all topics from the content of the program into educational process.

Practical classes on discipline are carried out at the clinical base of the department of propaedeutics of pediatrics and medical genetics (Lviv Regional Clinical Pediatric Hospital). Lectures are given in the conference hall of the same hospital. The content of lectures reflects the problematic issues of the relevant sections of propaedeutics of pediatrics. Duration of practical classes and lectures is 2 hours.

Practical classes are aimed at controlling the acquisition of theoretical material and the formation of practical skills, as well as the ability to analyze and apply the knowledge gained to solve practical problems. The main target of each practical class is the study of the physical and neuro-psychological development of children of different age groups; anatomical and physiological features of the child's organ systems; semiotics of syndromes, manifestation of disorders of different systems and the most common diseases of the child's organism, as well as the principles of rational nutrition. Means of control are MCQ, study cases; control of practical skills.

The following methodology for conducting practical classes is used:

1. Each class begins with 10-15 minutes test control in order to assess the initial level of knowledge and determine the degree of readiness of students to study.
2. Within 15-20 minutes the teacher explains and demonstrates the methodology of examination of a child, introduces students to the principles of organizing rational nutrition, etc.

3. Within 30-35 minutes students work with sick children independently, collect anamnesis, make examination, perform diagnostic and therapeutic manipulations, etc. During independent work, the teacher assists students and accentuates attention on the most important issues on this topic of practical classes.
4. Within 20-30 minutes students report about the results of their self-work. The teacher discusses, and explaning, emphasizes the peculiarities of different clinical cases. During the clinical examination, teacher controls the final level of knowledge of students.
5. After completing a practical class, teacher within 10-15 minutes summarise practical class, gives students the task for independent work, and offers a list of recommended literature for self-study.

8. Verification of learning outcomes

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.

Types of routine control:

- test tasks (MCQ)
- typical situational problems (study cases)
- evaluation of practical skills

Routine control

Code of studying results	Code od type of training	Method of verifying learning outcomes	Enrollment criteria
Kn-1, Com-1, AR-1 Kn-2, Com -2, AR-2 Kn-3, Com -3, AR-3 Kn-4, Com -4, AR-4 Kn-5, Com -5, AR-5 Kn-6, Com -6, AR-6 Kn-17, Com -17, AR-17	L-1	Control of knowledge of lecture material is carried out in practical classes, the topics of which correspond to the topic of the lecture. Routine control 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. 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. 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 study cases. 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	<u>Initial stage of the practical training:</u> 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. <u>The main stage of practical training:</u> The survey is rated 12, 8, 4, 0 points. <u>At the final stage of practical training:</u> 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” Self-work of the student is estimated as "Passed" / "Not
Kn-1, Com -1, AR-1 Kn-2, Com -2, AR-2 Kn-3, Com -3, AR-3 Kn-17, Com -17, AR-17	L-2		
Kn-1, Com -1, AR-1 Kn-2, Com -2, AR-2 Kn-3, Com -3, AR-3 Kn-4, Com -4, AR-4 Kn-5, Com -5, AR-5 Kn-6, Com -6, AR-6 Kn-17, Com -17, AR-17 Kn-21, Com -21, AR-21 Kn-24, Com -24, AR-24	L-3		
Kn-1, Sk-1, Com -1, AR-1 Kn-2, Sk-2, Com -2, AR-2 Kn-17,Sk-17, Com -17,AR-17	P-1		
Kn-1, Sk-1, Com -1, AR-1 Kn-2, Sk-2, Com -2, AR-2 Kn-3, Sk-3, Com -3, AR-3	P-2, P-3, P-4, P-5		
Kn-1, Sk-1, Com -1, AR-1 Kn-2, Sk-2, Com -2, AR-2 Kn-3, Sk-3, Com -3, AR-3 Kn-5,Sk-5, Com -5,AR-5 Kn-6, Sk-6, Com -6, AR-6	P-6		
Kn-1, Sk-1, Com -1, AR-1 Kn-2, Sk-2, Com -2, AR-2 Kn-3, Sk-3, Com -3, AR-3 Kn-17,Sk-17, Com -17,AR-17	P-7, SW-1, SW-2-4		

Kn-1, Sk-1, Com -1, AR-1 Kn-2, Sk-2, Com -2, AR-2 Kn-3, Sk-3, Com -3, AR-3 Kn-4, Sk-4, Com -4, AR-4 Kn-5, Sk-5, Com -5, AR-5 Kn-6, Sk-6, Com -6, AR-6 Kn-17, Sk-17, Com -17,AR-17 Kn-21, Sk-21,- Com 21,AR-21 Kn-24,Sk-24, Com -24, AR-24	П-8, SW-12	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	passed" in its implementation or failure of each semester results
Kn-1, Sk-1, Com -1, AR-1 Kn-2, Sk-2, Com -2, AR-2 Kn-3, Sk-3, Com -3, AR-3 Kn-4, Sk-4, Com -4, AR-4 Kn-5, Sk-5, Com -5, AR-5 Kn-6, Sk-6, Com -6, AR-6 Kn-17,Sk-17, Com -17,AR-17	SW-5 - 11		

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.

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	4- point scale	200- point scale	4- point scale	200- point scale	4- point scale	200- point scale
5.00	200	4.50	180	3.99	160	3.47	139
4.97	199	4.47	179	3.97	159	3.45	138
4.95	198	4.45	178	3.94	158	3.42	137
4.92	197	4.42	177	3.92	157	3.40	136
4.90	196	4.40	176	3.89	156	3.37	135
4.87	195	4.37	175	3.87	155	3.35	134
4.85	194	4.35	174	3.84	154	3.32	133
4.82	193	4.32	173	3.82	153	3.30	132
4.80	192	4.30	172	3.79	152	3.27	131
4.77	191	4.27	171	3.77	151	3.25	130
4.75	190	4.24	170	3.74	150	3.22	129
4.72	189	4.22	169	3.72	149	3.20	128

4.70	188
4.67	187
4.65	186
4.62	185
4.60	184
4.57	183
4.55	182
4.52	181

4.19	168
4.17	167
4.14	166
4.12	165
4.09	164
4.07	163
4.04	162
4.02	161

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

3.17	127
3.15	126
3.12	125
3.10	124
3.07	123
3.05	122
3.02	121
3	120
< 3	Not enough

Maximal score, which the student can get for the current study is 200.
Minimal score, which the student can get for the current study is 120.

Final control

General evaluation system	Educational activity during the semester / final control - 60% / 40% on a 200-point scale
Rating scales	Traditional 4-point scale, multi-point (200-point) scale, ECTS rating scale
Conditions of admission to the final control	The student attended all practical classes and received at least 120points for current study

Type of final control	Methods of final control	Criteria
Credit	All topics submitted for current control must be included. Grades from the 4-point scale are converted into points on a multi-point (200-point) scale in accordance with the Regulation "Criteria, rules and procedures for evaluating the results of student learning activities"	The maximum number of points that a student can score for the current academic activity in the study of the discipline is 200 points. The minimum number of points that a student must score for the current academic activity in the study of the discipline is 120 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).

The procedure for considering the appeal of a higher education applicant

In the case of disagreement of the student with the current evaluation, repassing of the topic is carried out in the presence of a commission consisting of three teachers, including head of the department. If the incident is not resolved, the information is submitted to the dean's office.

9. COURSE POLICY

The policy of the discipline is determined by the system of requirements for the student in the study of the discipline "Medical genetics" and is based on the principles of academic integrity. Students are explained the value of acquiring new knowledges, the academic norms that must be followed, why they are important, what is academic integrity, what are its values and functions, how students can contribute to its development by their actions; the essence, features and reasons of inadmissibility of academic plagiarism are explained, students of higher education are encouraged to perform educational tasks independently, to correctly refer to sources of information in case of borrowing of ideas, statements, information

Applicants of higher education must perfectly possess clinical thinking, fundamental and special knowledge and skills regarding the nature of hereditary human diseases, their etiology, pathogenesis, reasons for wide clinical polymorphism of etiologically uniform forms and genetic heterogeneity of clinically similar conditions; as well as methods of diagnosis of the most common forms of hereditary pathology and principles of interaction of the medical and genetic service with all services of health care practice.

The discipline "Medical Genetics" is compulsory for students of speciality 222 "Medicine". The student is obliged to fully master the knowledge, skills, practical skills and competencies in this discipline.

Policy compliance with the principles of academic integrity by applicants of higher education:

- the student's compliance with the student's code of ethics
- independent performance of educational tasks of current control without the use of external sources of information, except as permitted by the teacher, preparation of practical tasks during the class;
- write-offs during control are prohibited (including with the use of mobile devices);
- if student does not follow rules of academic integrity during practical classes, he/she receives a nonpositive mark ("2") and have to repass the topic;
- a report documenting a student's systematic violation of academic integrity is submitted to the dean's office.

Policy compliance with the principles and norms of ethics and deontology by applicants of higher education:

- actions in professional and educational situations from the standpoint of academic integrity and professional ethics and deontology;
- compliance with the rules of internal regulations of the clinical base of the department, to be tolerant, friendly and balanced in communication with students and teachers, patients, medical staff of health care institutions;
- awareness of the importance of examples of human behavior in accordance with the norms of academic integrity and medical ethics.

Policy of attendance for applicants of higher education:

- attendance at all classes is mandatory for the purpose of current and final control (except for serious reasons).

The policy of repassing topics and working off missed classes by applicants of higher education:

- passing of missed classes is conducted according to the schedule
- passing the topic of the lesson, for which the student received a negative mark, is carried out at a convenient time for the teacher and the student after the practical class, the maximum mark is "good"
- repassing the topic during the current training in order to increase the score is not allowed

10. LIST OF REFERENCE MATERIALS

Recommended literature

1. Essential Medical Genetics / M. Connor & M/ Ferguson-Smith Ed., 6th ed. – “Blackwell Science Ltd”. – 2011. – 344 p.
2. Georg F. Hoffmann, Johannes Zschocke. Vademecum Metabolicum, 2015.
3. Medical Genetics / Lynn Jorde, John Carey, Michael Bamshad // 6th Edition - September 27, 2019. – 352 p.
4. Wallace C.D., Brown M.D., Lott M.T. Mitochondrial genetics. – 2007
5. Zhadanov S.I., Grechanina E.Ya., Grechanina Yu.B., Gusar V.A., Fedoseeva N.P., Lebon S., Münnich A., Schurr T.G. «Fatal manifestation of a de novo ND5 mutation: Insights into the pathogenic mechanisms of mtDNA ND5 gene defects». Mitochondrion. 2007. – P.260-266.
6. McKusick V.A. Mendelian inheritance in man. 10-th ed. v.l,2.Johris Hopkins Univ.Press. 2008.

Information sources

1. http://www.ncbi.nlm.nih.gov/omim
2. Rehm, H.L. (2013 April). Disease-targeted sequencing: a cornerstone in the clinic. Nature Reviews Genetics. Available online at https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3786217/. Accessed October 2018.
3. Katsanis, S. H. and Katsanis N. (2013 June). Molecular genetic testing and the future of clinical genomics. National Review of Genetics. Available online at https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4461364/. Accessed October 2017.
Behjati, S. and Tarpey P.S. (2013 December). What is next generation sequencing? Archives of Disease in Childhood. Available online at https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3841808/. Accessed October 2018.

4. Shen T., et al. (2015 June 17). Clinical applications of next generation sequencing in cancer: from panels, to exomes, to genomes. Frontiers in Genetics. Available online at https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4469892/. Accessed October 2018.
5. Moynihan, T. (2015 June 30). Gene expression profiling for breast cancer: What is it? Mayo Clinic. Available online at https://www.mayoclinic.org/diseases-conditions/breast-cancer/expert-answers/gene-expression-profiling/faq-20058144. Accessed October 2018
6. (2015 August 27). DNA microarray technology. National Human Genome Research Institute. Available online at https://www.genome.gov/10000533/dna-microarray-technology/. Accessed October 2018.
7. (Updated 2015 December 18). DNA sequencing. National Human Genome Research Institute. Available online at https://www.genome.gov/10001177/dna-sequencing-fact-sheet/. Accessed October 2018.
8. Benowitz, S. (2016 March 29). New study challenges gold standard for validating DNA sequencing results. National Human Genome Research Institute. Available online at https://www.genome.gov/27564480/2016-news-feature-new-study-challenges-gold-standard-for-validating-dna-sequencing-results/. Accessed October 2018.
9. Hamilton, A. et al. (2016 May 10). Concordance between whole-exome sequencing and clinical Sanger sequencing: implications for patient care. Molecular Genetics and Genomic Medicine. Available online at http://onlinelibrary.wiley.com/doi/10.1002/mgg3.223/full. Accessed October 2018.
10. Sabour L., et al. (2017 April). Clinical applications of next-generation sequencing in cancer diagnosis. Pathology and Oncology Research. Available online at https://link.springer.com/article/10.1007/s12253-016-0124-z. Accessed October 2018.
11. (© 2017). What is diagnostic testing? Genes in Life. Available online at http://www.genesinlife.org/testing-services/testing-genetic-conditions/diagnostic-testing. Accessed October 2018.
12. (© 2017). What is genotyping and expression profiling? Genotyping and Coriell Institute for Medical Research. Available online at https://www.coriell.org/research-services/genotyping-microarray/what-is-genotyping-and-expression-profiling. Accessed October 2018.
13. (August 27, 2015) National Human Genome Research Institute, DNA Microarray. Available online https://www.genome.gov/10000533/dna-microarray-technology/ Accessed October 2018.

11. EQUIPMENT, MATERIAL AND TECHNICAL SUPPORT OF THE DISCIPLINE

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.

12. ADDITIONAL INFORMATION

During the practical training the student must be dressed in a medical suite and cap, have medical shoes and a medical mask, because studying process takes place in the hospital. During study it is necessary to have computer equipment (with access to the global network) and office equipment for communication with teachers and preparation (printing) of reports, presentations.

Information materials related to the educational and organizational process (thematic plans, class schedule, schedules of consultations and repassings of missed classes) are available on the website of the Department of Propaedeutics Pediatrics and Medical Genetics:
https://new.meduniv.lviv.ua/kafedry/kafedra-propedevtyky-pediatriyi-ta-medychnoyi-genetyky/

Methodical guides for preparation for practical classes, independent work, self-control, abstracts of lectures are available on the MISA platform in the section "Distance study" on the website of Danylo Halytskyi Lviv National Medical University

http://misa.meduniv.lviv.ua/login/index.php

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