Ministry of Healthcare of Ukraine
Poltava State Medical University

Department of Nervous Diseases

# SYLLABUS

# INTERNAL MEDICINE, INCLUDING ENDOCRINOLOGY, MEDICAL GENETICS

(title of the academic discipline)

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|---|---|
| academic and professional level | the second (master's) level of higher education |
| field of knowledge | 22 «Healthcare» |
| specialty | 222 «Medicine» |
| academic qualification | Master of Medicine |
| professional qualification | Doctor |
| academic and professional program | «Medicine» |
| mode of study | full-time |
| course(s) and semester(s) of study of the discipline | 4 course, VII, VIII semester |

## Module 3. MEDICAL GENETICS

«**RESOLVED**»
at the meeting of the
Department of Nervous Diseases

Head of the Department _____ Mykhailo DELVA

Minutes as «____»_____ 2023, №___

Poltava – 2024

## INFORMATION ABOUT LECTURERS WHO DELIVER THE ACADEMIC DISCIPLINE

| | |
|---|---|
| Surname, name of teachers, scientific degree, academic rank | Hryn Kateryna, PhD, docent |
| Teachers' profile | https://ndiseases.pdmu.edu.ua/ |
| Contact number | Neurological department of Poltava regional hospital named after M.V. Sklyfosovsky. – 23 Shevcenko str., tel. (0532) 56-42-37, (0532) 52-49-05. |
| E-mail: | neurology@pdmu.edu.ua |
| Department's website | https://ndiseases.pdmu.edu.ua/ |

## MAIN CHARACTERISTICS OF THE ACADEMIC DISCIPLINE

**The volume of the academic discipline – module 3. Medical Genetics**

Number of credits / hours − 1,0/30 among them:

Lectures (hours) − 0

Workshops (hours) − 20

Individual work (hours) − 10

Type of control – Final modular control (FMC).

**The policy of the academic discipline**

The policy of academic discipline is based on the conscious and conscientious performance of higher education students of their duties, compliance with the general rules and norms of behavior accepted in society.

During organizing of educational process at PSMU, teachers and higher education seekers act according to:

- «Regulations on the organization of the educational process at Poltava State Medical University»;
- «Regulations on the academic integrity of applicants for higher education and employees of Poltava State Medical University»;
- «Rules of Procedure for students of Poltava State Medical University»;
- «Regulations on the organization and methodology of assessment of educational activities of applicants for higher education at Poltava State Medical University»;
- «Regulations on the organization of independent work of students at Poltava State Medical University»;
- «Regulations on the completion of missed classes and unsatisfactory grades by applicants for higher education of Poltava State Medical University»;
- «Regulations on the procedure for forming individual educational trajectories by students of Poltava State Medical University»;
- «Regulations on the rating of applicants for higher education of Poltava State Medical University»;
- «Regulations on the procedure for re-enrollment of academic disciplines and determination of academic difference»;
- «Regulations on the appeal of the results of the final control of knowledge of applicants for higher education», etc.

You can get acquainted with the above provisions at https://www.pdmu.edu.ua/n-process/department-npr/normativni-dokumenti

While studying at the Department of Nervous Diseases with Neurosurgery and Medical Genetics, higher education seeker must follow the rules that are adopted by the Academy. Higher education seeker, as a future doctor, must have a high level of culture of behavior, behave with dignity, tact, maintain endurance and self-control both during training in practice and when dealing with patients in the department.

Higher education seeker is required to attend classes in time, according to the class schedule. It is not allowed to violate the schedule of the educational process do not fulfill of the curriculum and individual curriculum, to be late for classes, to miss classes without good reasons. Missed classes must be reworked for all higher education seekers, regardless of the sources of funding for their education. Higher education seeker at Poltava State Medical University work off missed classes regardless of the reason for absence, in electronic classes № 1, № 2, № 3, according to the schedule, in accordance with the «Regulations on working off missed classes and unsatisfactory marks by higher education seekers at Poltava State Medical University».

During their stay at the department and clinical bases, participants of the educational process must comply with the requirements for the appearance of persons who work and study at the academy, be dressed in appropriate medical uniforms.

Higher education seekers are prohibited from leaving the classroom without the teacher's permission, using a mobile phone and other means of communication and receiving information without the teacher's permission, engaging in extraneous

activities, distracting other higher education seekers and disturbing the teacher. During the stay at the clinical base and in the surrounding areas, participants in the educational process are prohibited from smoking.

During the study of the discipline, higher education seekers are required to adhere to the rules of academic integrity, which provides self-performance of educational tasks, tasks for current and for final control; references to sources of information in the case of the use of ideas, developments, statements, information; compliance with the law on copyright and related rights; providing reliable information about the results of their own educational (scientific, creative) activities, used research methods and sources of information.

**The academic discipline description**

**Medical genetics** is a science that studies the role of heredity and variability in different populations, features of manifestation and development of normal and pathological signs, patterns of transmission of hereditary diseases from generation to generation, dependence of diseases on genetic or epigenetic abnormalities, develops diagnostic methods, treatment, prevention of hereditary pathology, including diseases with hereditary predisposition.

The task of medical genetics is to identify, study, prevent and treat hereditary diseases, to develop ways to prevent the impact of negative environmental factors on human heredity.

The subject of study of the academic discipline «Internal Medicine, including Endocrinology, Medical Genetics» is a clinical genetics that studies patterns of heredity and variability in humans for all levels of its organization and being: molecular, cellular, organismic, population, biochronological, biogeochemical. Medical genetics studies the place of heredity in human pathology, patterns of vertical transmission of hereditary diseases, develops methods of diagnosis, treatment, prevention of hereditary pathology, including diseases with hereditary predisposition.

The task of modern medicine is a gradual transition from the field of treatment of patients to the prevention of disease and public health, and knowledge of the basics of medical genetics requires a doctor in the diagnosis, treatment of diseases and preventive measures.

It is important to create in applicants for higher education an idea of the patterns of inheritance and variability of traits in relation to human pathology; causes of hereditary human diseases; the nature of their inheritance in families; distribution in populations; basic principles of diagnosis, treatment and prevention of hereditary human pathology.

**Prerequisites and post-requisites of the discipline (interdisciplinary communication).**

Medical genetics as an academic discipline:

a) is based on the knowledge, received by applicants for higher education of medical biology and genetics, medical and biological physics, medical chemistry, biological and bioorganic chemistry, morphological disciplines, normal and

pathological physiology, propedeutics of internal and childhood diseases and integrates with these disciplines;

b) lays basis of modern diagnostic technologies study which are used not only in diagnosis of hereditary diseases, but also in general clinical practice, which involves the integration of teaching with different disciplines and the formation of skills to apply knowledge of modern methods of genetic diagnostics in the process of further learning and in the professional activity;

c) introduces understanding of modern features of Single-Gene Disorders and Chromosomal Diseases, as well as common ailments that occur against the background of hereditary predisposition and require the integration of classical clinical concepts and modern high technology.

**The purpose and tasks of the discipline:**

The purpose of academic discipline «Internal medicine, including endocrinology, medical genetics, module 3. Medical genetics» is the study of a phenomena of heredity and variability among populations, the features of manifestation and development of normal and pathological features, dependence of diseases on genetic or epigenetic abnormalities.

The main tasks of the study of Medical Genetics are detection, study, prevention and treatment of hereditary diseases, development of ways to prevent the influence of external negative environmental factors on human heredity.

**Competences and learning outcomes, the formation of which is facilitated by the discipline «Internal medicine, including endocrinology, medical genetics, module 3. Medical genetics» (integral, general, special)**

*Integral competence:*

The ability to solve complex problems, including research and innovation in the field of medicine. The ability to continue study with a high degree of autonomy.

*General competencies:*

1. Social and legal competences:

1.1 awareness of equal opportunities and gender issues;

1.2 the ability to exercise their rights and obligations as a member of society, to realize the values of civil (free democratic) society and the need for its sustainable development, supremacy of law, rights and freedoms of a person and citizen;

1.3 the ability to preserve and increase moral, cultural, scientific values and achievements of society based on understanding history and patterns of development of the subject area, its place in the general system of knowledge about nature and development of the society, machinery and technology, use different types and forms of motor activity for the active recreation and healthy living;

2. Prognostic competencies:

2.1 ability to abstract thinking, analysis and synthesis;

2.2 the ability to learn and master modern knowledge;

2.3 knowledge and understanding of the subject area and understanding of professional activity;

2.4 ability to make rational decisions;
2.5 ability to work in a team;
2.6 ability to use information and communication technologies;
2.7 ability to search, process and analyze information from various sources.
3. Communicative competences:
3.1 ability to interpersonal interaction;
3.2 ability to communicate in a foreign language.
4. Management competences:
4.1 ability to apply knowledge in practical situations;
4.2 ability to adapt and act in a new situation;
4.3 certainty and perseverance regarding the tasks and responsibilities taken.

*Special (professional, subject) competencies:*

1. The ability to collect medical information about the patient and analyze clinical data.
2. The ability to determine the required list of laboratory and instrumental studies and evaluate their findings.
3. The ability to establish a preliminary and clinical diagnosis of the disease.
4. The ability to determine the necessary mode of work and rest in the treatment and prevention of diseases.
5. The ability to determine the nature of nutrition in the treatment and prevention of diseases.
6. The ability to determine the principles and nature of treatment and prevention of diseases.
7. The ability to diagnose emergencies.
8. The ability to determine tactics and provide emergency medical care.
9. The ability to perform medical manipulations.
10. The ability to untangle medical problems in a new or unfamiliar environment in the presence of incomplete or limited information, taking into account aspects of social and ethical responsibility.
11. The ability to determine the tactics of management of physiological pregnancy, physiological labor and the postpartum period. To conduct consultations on family planning and methods of contraception
12. The ability to conduct sanitary and hygienic and preventive measures.
13. The ability to make medical records, including electronic forms.
14. The ability to assess the impact of the environment, socioeconomic, biological determinants on the health of the individual, family, population.
15. The ability to develop and implement scientific and applied projects in the field of public health.
16. Compliance with ethical principles when working with patients, their relatives, laboratory animals.
17. Compliance with professional and academic integrity, responsibility for the reliability of scientific results obtained.

**Program learning outcomes, the formation of which is facilitated by the discipline «Internal medicine, including endocrinology, medical genetics, module 3. Medical genetics»**

1. Have a thorough knowledge of the structure of professional activity. Be able to carry out professional activities that require updating and integrating knowledge. Be responsible for professional development, the ability to further professional training with a high level of autonomy.
2. Specialized conceptual knowledge, including scientific achievements in the field of public health and is the basis for research, critical understanding of problems in the field of medicine and related interdisciplinary problems.
3. Distinguish and identify major 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 on a human being, the organs and systems of the body, establish a preliminary clinical diagnosis of the disease (according to list 2).
4. Collect complaints, history of life and disease, evaluate the psychomotor and physical development of the patient, the state of organs and systems of the body, and estimate the information on the diagnosis (according to the list 4), taking into account the age of the patient.
5. To establish the final clinical diagnosis by making a rational decision and analyzing the obtained subjective and objective data of clinical, additional examination, differential diagnosis, adhering to the relevant ethical and legal norms, under the supervision of a physician in a health care facility (according to list 2).
6. Prescribe and analyze additional (mandatory and optional) methods of examination (laboratory, functional and / or instrumental) (according to list 4), patients with diseases of organs and body systems for differential diagnosis of diseases (according to list 2).
7. Determine the main clinical syndrome or what determines the severity of the condition of the victim (according to the list 3) by making a reasonable decision and assessing the human condition in any circumstances (in the conditions of a health care facility, outside) including in an emergency and hostilities, in the battlefield, in conditions of lack of information and limited time.
8. Determine the nature and principles of treatment of patients (conservative, operative) with diseases (according to list 2), taking into account the age of the patient, in the conditions of the health care facility, outside and at the stages of medical evacuation, including on the battlefield, on the basis of the preliminary clinical diagnosis, adhering to the relevant ethical and legal norms, by making a reasonable decision according to existing algorithms and standard schemes, if it is necessary to expand the standard scheme, be able to justify personalized recommendations under the supervision of a physician in a medical facility.
9. Determine the necessary mode of work, rest and nutrition on the basis of the final clinical diagnosis, adhering to the relevant ethical and legal norms, by making a rational decision on existing algorithms and standard schemes.
10. To form rational medical routes of patients; organize interaction with colleagues in their own and other facilities, organizations and institutions; apply tools for promoting medical services on the market, based on the analysis of the needs of the

population, in the conditions of functioning of the health care facilities, its unit, in a competitive environment.

11. Perform medical manipulations (according to the list of 5) in a medical facility, at home or at the enterprise on the basis of a preliminary clinical diagnosis and / or indicators of the patient's condition by making a rational decision, adhering to the relevant ethical and legal norms.

12. To find the necessary information in the professional literature and databases of other sources, to analyze, evaluate and apply this information.

13. Apply the state-of-the-art digital technologies, specialized software, statistical methods of data analysis to solve complex health problems.

14. To convey clearly and unequivocally the own knowledge, conclusions and arguments on health problems and related issues to specialists and non-professionals.

15. Freely communicate in the national and English language, both orally and in writing to discuss professional activities, research and projects. Use international Greek-Latin terms, abbreviations and clichés in professional oral and written speech.

**Learning outcomes of the academic discipline:**

upon completing their study in the academic discipline of module 3. Medical Genetics higher education seekers must

**to know:**

– Algorithm for examination of the patient and his family members for hereditary diseases.

– Methods of medical genetics, their advantages and indications for implementation.

– Chromosome staining methods.

– Algorithm for compiling the pedigree and legend to the pedigree.

– Methodology of clinical and genealogical analysis of the family tree.

– Methodology of working with diagnostic catalogs.

– Algorithms for conducting syndromic analysis in the process of diagnosing hereditary pathology.

– Types of inheritance.

– The concept of morphogenetic variants of development.

– Types of violations in the chromosomal set: structural, numerical.

– Classification of chromosomal diseases depending on the changes, structure, number or violation of the ploidy of the chromosomal set.

– The concept of "mosaicism".

– To explain the phenomena of chromosomal aberrations.

– Principles of organization of screening programs.

– Criteria for mass and selective screening for hereditary metabolic diseases (HMD).

– Basic research methods in case of suspicion of HMD.

– Schemes and algorithms for examination of patients with suspected HMD of amino acids, carbohydrates, connective tissue, organic aciduria.

– Indications for carrying out general metabolic screening tests of urine.

– Mechanisms of acute metabolic disorders in the neonatal period.

– Indications for the analysis of organic acids.

– Indications for the study of the exchange of connective tissue.
– Mechanisms of ketosis and lactic acidosis in patients with organic aciduria.
– Clinical value of screening programs in early diagnosis of HMD.
– The program of biochemical diagnosis of HMD with an acute course.
– The value of routine biochemical studies in the diagnosis of HMD.
– Mechanisms of hypoglycemia in patients with organic aciduria.
– Basic research methods for fatty acid metabolism disorders.
– Criteria for selection of high genetic risk groups for the development of HMD.
– The structure and function of the nuclear and mitochondrial genome.
– Modern possibilities of prenatal diagnostics.
– Methods of prenatal diagnosis.
– Indications for invasive prenatal diagnosis.
– Deadlines for screening pregnant women.
– Indications for pregnancy elimination.

**be able to:**

– Apply a systematic approach when examining the patient and his family members.
– Determine the leading clinical symptom complex when assessing the phenotype of the proband and his family.
– Compile an algorithm for the examination of a patient with suspected hereditary pathology.
– Assess the phenotype of the proband and his family members.
– Carry out a syndromic analysis.
– Work with diagnostic catalogs.
– Compile and analyze the pedigree and legend to the pedigree.
– Assess the nature of inheritance of a phenotypic trait or disease based on a given pedigree.
– Draw a diagram of the structure of nucleotides, internucleotide and interchain bonds.
– Draw a diagram of the periods of the cell cycle.
– Draw a diagram of mitosis.
– Draw a diagram of meiosis.
– Interpret kariograms in normal and pathological conditions.
– To interpret the mechanisms of division of somatic and germ cells.
– Explain the methods of obtaining preparations of mitotic chromosomes.
– Draw a diagram of the occurrence of trisomies. Draw a diagram of the occurrence of monosomies.
– Draw a diagram of the occurrence of structural chromosomal rearrangements.
– Develop an algorithm for molecular cytogenetic research.
– Analyze the chromatogram.
– Interpret the results of thin-layer chromatography of amino acids and carbohydrates of blood and urine.
– Interpret the results of general metabolic screening urine tests.
– Illustrate with examples the importance of biochemical studies in clarifying the diagnosis of SHO.
– Interpret the results of the study of the exchange of connective tissue.

– Draw a diagram of phenylalanine exchange.
– Draw a diagram of methionine exchange.
– Analyze the results of PCR analysis.
– Interpret the results of DNA diagnostics of monogenic and infectious diseases.
– Analyze sonograms.
– Compile an algorithm for cytogenetic research of chorion cells, blood lymphocytes, and amniocytes.
– Analyze the results of biochemical screening.

**Thematic plan of lectures (by modules) with the indication of the basic questions considered at lectures –** is not provided by the working curriculum

**Thematic plan of seminars by modules and content modules with the indication of the main issues considered at the seminar –** is not provided by the working curriculum

**Thematic plan of practical classes by modules and content modules, specifying the basic issues, which are considered at the practical class**

| № | TOPIC | Number of hours |
|---|---|---|
| **Content module 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.**<br>1. Molecular basis of heredity. Types of trait inheritance.<br>2. The place of hereditary pathology in the structure of morbidity and mortality.<br>3. Types of hereditary variability<br>4. Pathogenesis of hereditary diseases.<br>5. Classification of hereditary diseases.<br>6. The main tasks of medical genetics.<br>7. Chromosomal, genomic and gene mutations, their role in human pathology.<br>8. Lethal and sublethal mutations. Causes of mutations.<br>9. Spontaneous and induced human mutations, frequency of their occurrence.<br>10. Physical, chemical and biological mutagenesis. Medicinal mutagenesis, teratogenesis, carcinogenesis. | 2 |
| **Content module 2. Methods of medical genetics.** | | |
| 2 | **Clinical and genealogical method. Cytogenetic and molecular genetic techniques. Biochemical techniques.**<br>1.Stages of conduction of clinical and and genealogical examination.<br>2. Place of clinical and genealogical method in clinical practice.<br>3. Genealogical, twin and population- based and statistical methods in clinical and genetic analysis of multifactorial diseases.<br>4. Application field of cytogenetic methods.<br>5. Alternative study methods of cytogenetic.<br>6. Place of the cytogenetic method in clinical practice. | 2 |

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| | 7. Methods of DNA-diagnostics, potential for their use.<br>8.Availabilities of molecular genetic techniques in the diagnosis of hereditary diseases.<br>9. Place of biochemical techniques in the diagnosis of hereditary metabolic diseases and multifactorial diseases.<br>10. Indications for biochemical screen for the diagnosis of hereditary diseases. | |
| **Content module 3. Propaedeutics of hereditary pathology.** | | |
| 3 | **Semiotics of hereditary diseases. Features of manifestations of hereditary diseases. Morphogenetic variants of development. Abnormalities of development.**<br>1. Semiotics of hereditary pathology.<br>2. Phenotype, its features.<br>3. Pleiotropy, pleiotropy of genes and numerous affection in the case of hereditary pathology.<br>4. Morphogenetic variants of development, their genesis, postnatal modification.<br>5. Primary and secondary abnormalities.<br>6. Isolated, systemic and numerous congenital disorders.<br>7. Consistency of the nature of disorders with the stages of ontogenesis (gameto-, blasto-, embryo- and fetopathy). | 2 |
| **Content module 4. Single-Gene Disorders.** | | |
| 4 | **General characteristics of Single-Gene Disorders. Clinics and genetics of certain forms of Single-Gene Disorders and Epigenetic Diseases**.<br>1. Etiology and pathogenesis of monogenic diseases, frequency in the population.<br>2. Classification of monogenic diseases and syndromes. Genetic heterogeneity.<br>3. Features of the clinical picture of monogenic diseases.<br>4. Catalog of genes and genetic diseases by V. McKusick.<br>5. Monogenic diseases and syndromes with an autosomal dominant type of inheritance: achondroplasia, Marfan syndrome, acrocephalosyndactyly (Aper syndrome), Ehlers-Danlo syndrome. Clinic, diagnosis, treatment, prenatal diagnosis, medical and genetic counseling, determination of genetic risk.<br>6. Monogenic diseases and syndromes with an autosomal recessive type of inheritance: phenylketonuria, cystic fibrosis, galactosemia, hypothyroidism, adrenogenital syndrome.<br>7. Monogenic diseases and syndromes with an X-chromosome-linked type of inheritance: Duchenne-Becker muscular dystrophy, fragile X-chromosome syndrome, phosphate diabetes, hemophilia.<br>8. Diagnosis of monogenic diseases. Biochemical methods. Selective and mass screening.<br>9. Principles of treatment of monogenic diseases: symptomatic, pathogenetic and etiological. | 2 |
| 5 | **Metabolic hereditary diseases. Principles of treatment of hereditary diseases, rehabilitation and social adaptation.**<br>1. Hereditary metabolic diseases, their classification. Clinical symptoms indicating a hereditary metabolic disorder.<br>2. Accumulation diseases.<br>3. Mucopolysaccharidoses. | 2 |

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| | 4. Sphingolipidosis.<br>5. Peroxisomal diseases.<br>6. Clinic and genetics of individual forms of hereditary metabolic diseases with different types of inheritance: homocystinuria, glycogenoses, Gaucher disease, Niemann-Pick disease.<br>7. Their frequency in the population, clinical forms and variants, types of mutations, pathogenesis, typical clinical picture, paraclinical and laboratory methods of diagnosis, treatment, prognosis, rehabilitation, social adaptation.<br>8. Symptomatic and pathogenetic therapy of hereditary metabolic diseases.<br>9. Principles of pathogenetic treatment as the main method of therapy of hereditary diseases. | |
| **Content module 5. Chromosomal diseases** | | |
| 6 | **General characteristics of chromosomal diseases. Clinic of main forms of chromosomal diseases.**<br>1. Chromosomal diseases. Definition of the concept. Etiology, pathogenesis and classification.<br>2. General characteristics of chromosomal diseases.<br>3. Clinical and genetic characteristics of Patau syndrome.<br>4. Clinical and genetic characteristics of Edwards syndrome.<br>5. Clinical and genetic characteristics of Down's syndrome.<br>6. Clinical and genetic characteristics of trisomy 22.<br>7. Clinical and genetic characteristics of Turner's syndrome.<br>8. Clinical and genetic characteristics of polysomies according to sex chromosomes.<br>9. Clinical and genetic characteristics of partial aneuploidy syndromes.<br>10. Clinical and genetic characteristics of microcytogenetic syndromes.<br>11. Factors of increased risk of having children with chromosomal diseases. | 2 |
| **Content module 6. Mitochondrial diseases** | | |
| 7 | **General characteristics of mitochondrial pathology. Clinic, diagnosis, treatment.**<br>1. General characteristics of mitochondrial pathology. Classification of mitochondrial diseases.<br>2. Clinic, genetics, diagnosis, therapy of MERRF, MELAS syndromes.<br>3. Clinic, genetics, diagnosis, therapy of Leber, Cairns-Sayre syndromes, retinitis pigmentosa, Pearson syndrome.<br>4. Mitochondrial diseases caused by nuclear DNA mutations.<br>5. Diseases associated with respiratory chain defects.<br>6. Diseases associated with disturbances in the metabolism of lactic and pyruvic acids.<br>7. Diseases caused by defects in beta-oxidation of fatty acids.<br>8. General principles of diagnosis and treatment of mitochondrial pathology. | 2 |
| **Content module 7. Diseases with hereditary predisposition** | | |
| 8 | **General characteristics of multifactorial diseases. Determination of genetic predisposition. Preventive measures.**<br>1. The definition of multifactorial diseases.<br>2. General clinical characteristics of multifactorial diseases. | 2 |

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| | 3. Classification of multifactorial diseases.<br>4. Methods of study of multifactorial diseases.<br>5. Models of inheritance of multifactorial diseases.<br>6. The role of heredity in the development of coronary heart disease.<br>7. The role of heredity in the pathogenesis of diabetes.<br>8. The role of heredity in the development of peptic ulcer.<br>9. The role of heredity in the development of bronchial asthma.<br>10. Medical and genetic counselling in multifactorial pathology. | |
| **Content module 8. Prevention of hereditary pathology. Medical and genetic counseling and prenatal diagnosis.** | | |
| 9 | **Levels and ways of prevention of hereditary diseases. Medical and genetic counselling. Prenatal diagnosis. Screening programs.**<br>1. Medical and genetic counselling, the main stages of its conduction<br>2. The indications for medical and genetic counselling.<br>3. The definitions and requirements for prenatal diagnosis.<br>4. Mass diagnostics with sifting programs.<br>5. Noninvasive prenatal testing: ultrasound examination – indications, terms of conduction.<br>6. Invasive diagnostic procedures: fetoscopy; amniocentesis; chorionic biopsy; cordocentesis; placental biopsy; skin biopsy; muscle biopsy.<br>7. Indirect techniques of prenatal diagnosis: obstetric and gynecological; medical genetics (genealogical, cytogenetic, molecular genetics); microbiological; biochemical screening tests).<br>8. Methods of prevention of hereditary diseases. | 2 |
| | **Final modular control (FMC)** | **2** |
| | **Total** | **20** |

## Independent work

| № | Theme | Number of hours |
|---|---|---|
| 1 | Independent study of themes that are included into the class hours | 7 |
| 2 | Independent study of themes that are not included into the class hours | |
| 2.2 | **Systemic skeletal dysplasia**<br>1. Classification of systemic skeletal dysplasias (SSD).<br>2. Clinic, genetics, diagnosis of Jeun's syndrome, diastrophic dysplasia, achondroplasia, hypochondroplasia, imperfect osteogenesis, hypophosphatasia.<br>3. Prenatal diagnosis of SSD. Treatment and medical genetic counseling. | 1 |
| | **Preparate to FMC** | 2 |
| | **Total** | 10 |

## Individual tasks

1. Report at scientific seminars of the department.
2. Writing and presenting an essay on the topics of practical classes (free form).
3. Preparation of a literature review on clinical cases that have difficulties in differential diagnosis and/or treatment.
4. Selection of visual materials from sections of content modules.

5. Writing theses, articles.

6. Development of an algorithm for examining the family of a group of high genetic risk.

7. Participation in interuniversity Olympiads.

**The list of theoretical questions for preparation of applicants for the final modular control**

Content module 1. Heredity and pathology. The role of heredity in human pathology.

1. Subject and tasks of medical genetics. The value of genetics for medicine.

2. Frequency of congenital and hereditary pathology in different periods of ontogenesis.

3. Specific weight of congenital and hereditary pathology in the structure of morbidity and mortality.

4. The role of heredity, variability and environment in the development of pathology.

5. Classification of hereditary pathology.

Content module 2. Methods of medical genetics.

1. Role of paraclinical diagnostic techniques of congenital and hereditary pathology.

2. Cytogenetics and molecular cytogenetics techniques. Methods. Indications for cytogenic researches.

3. Clinical and genealogical method.

4. Methodology of lineage assembly.

5. Types of inheritance.

6. Biochemical techniques. Methods. Indications for diagnostic techniques.

7. Molecular genetic techniques. Methods. Indications and possibilities of the technique.

Content module 3. Propaedeutics of hereditary pathology.

1. Semiotics of hereditary diseases.

2. Features of clinical manifestations of congenital and hereditary pathology.

3. General principles of clinical diagnosis of congenital and hereditary pathology.

4. Features of inspection and physical examination of the patient and family members.

5. Congenital anomaly.

6. Congenital morphogenetic variants.

7. Syndromological approach in the diagnosis of congenital and hereditary pathology.

Content module 4. Single-Gene Disorders.

1. Monogenic diseases. Definition of the concept. Etiology and classification.

2. General regularities of the pathogenesis of monogenic pathology.

3. Genetic heterogeneity of monogenic diseases.

4. Genomic imprinting. Definition of the concept.

5. Clinic, genetics and diagnosis of neurofibromatosis.

6. Clinic, genetics and diagnosis of congenital hypothyroidism.

7. Clinic, genetics and diagnosis of phenylketonuria.

8. Clinic, genetics and diagnosis of cystic fibrosis.

9. Clinic, genetics and diagnosis of Marfan syndrome.

10. Clinic, genetics and diagnosis of homocystinuria.

11. Clinic, genetics and diagnosis of Ehlers-Danlo syndrome.

12. Modern classification of hereditary metabolic diseases (HMD).

13. Glycogenoses: frequency in the population, classification, clinical forms, methods of diagnosis and treatment, prognosis, rehabilitation, social adaptation.

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

Content module 5. Chromosomal diseases

1. Chromosomal diseases. Definition of the concept. Etiology, pathogenesis and classification.

2. General characteristics of chromosomal diseases.

3. Clinical and genetic characteristics of Patau syndrome.

4. Clinical and genetic characteristics of Edwards syndrome.

5. Clinical and genetic characteristics of Down's syndrome.

6. Clinical and genetic characteristics of trisomy 22.

7. Clinical and genetic characteristics of Turner's syndrome.

8. Clinical and genetic characteristics of polysomies according to sex chromosomes.

9. Clinical and genetic characteristics of partial aneuploidy syndromes.

10. Clinical and genetic characteristics of microcytogenetic syndromes.

11. Factors of increased risk of having children with chromosomal diseases.

Content module 6. Mitochondrial diseases

1. General characteristics of mitochondrial pathology. Mitochondrial inheritance.

2. Classification of mitochondrial diseases.

3. General principles of diagnosis and treatment of mitochondrial pathology.

4. Mitochondrial diseases caused by mitochondrial DNA mutations.

5. Clinic, genetics, diagnosis, therapy of Cairns-Sayre syndrome.

6. Clinic, genetics, diagnosis, therapy of MELAS syndrome.

7. Clinic, genetics, diagnosis, therapy of MERRF syndrome.

8. Clinic, genetics, diagnosis, therapy of Leber syndrome.

9. Clinic, genetics, diagnosis, therapy of Pearson's syndrome.

10. Mitochondrial diseases caused by nuclear DNA mutations.

Content module 7. Diseases with hereditary predisposition

1. Diseases with hereditary predisposition. Definition of the concept. General characteristics.

2. Mechanisms of disease development with hereditary predisposition.

3. The value of hereditary predisposition in general human pathology.

4. Hereditary pathological reactions to the action of external factors.

Content module 8. Prevention of hereditary pathology. Medical and genetic counseling and prenatal diagnosis.

1. Prevention of congenital and hereditary pathology. Types and levels of prevention.

2. Issues of family planning and preconception prevention.
3. Medical genetic counseling (MGC).
4. General provisions and indications for the MGK.
5. Prenatal diagnosis (PD). General questions. Indication. Deadlines.
6. Mass and selective ultrasound screening of pregnant women, timing, indications.
7. Non-invasive PD methods. Methods. Indication. Deadlines. Possibilities of the method.
8. Invasive PD methods. Methods. Indication. Deadlines. Possibilities of the method. Contraindication. Possible complications.
9. Preclinical diagnosis and preventive treatment.
10. Screening programs. Mass and selective screening programs.
11. Genetic monitoring of congenital and hereditary pathology.

**The list of practical skills for final module control:**

1. Determine the risk groups for the development of hereditary diseases.
2. Determine the algorithm of examination of patients with high genetic risk for the development of hereditary diseases.
3. Examine the patient with hereditary pathology, recognize the general manifestations of hereditary pathology, diagnose congenital morphogenetic variants, and using correcty appropriate terminology during description of clinical picture and phenotype of a patient.
4. Collect the anamnestic data and genealogical information, make up pedigree, present it in a graphical form and analyze the type of disease inheritance or disease in the family.
5. Choose from the patient population for cytogenetic, special biochemical and molecular genetic studies.
6. Define the provisional diagnosis of chromosomal pathology and some of the most common Single-Gene Disorders and syndromes; determine the need for additional examination, including specific genetic techniques.
7. Identify the individuals with increased risk of multifactorial disease.
8. Use clinical and genealogical method to evaluate the adverse effects of environmental factors.
9. Use methods of medical genetics for organization of observation (monitoring) of the long-term effects of environmental impacts.
10. Carry out the prophylactic measures focused on prevention of occurrence of hereditary and congenital diseases.
11. Carry out the preventive measures focused on the decrease of frequency of the most common diseases of multifactorial nature on the basis of genetic approaches.

**The form of summative assessment of academic achievement** − final modular control

**The system of continuous and final control**

The current control of educational activity is assessed on a traditional 4-point scale.

**Teaching methods.**

1. Verbal (lecture, explanation, communication, conversation, instruction)
2. Visual (observation, illustration, demonstration)
3. Practical (practice for developing skills)
4. Self-directed work of higher education seekers on comprehension and assimilation of new material
5. Thematic discussions
6. Brainstorming
7. Round table
8. Analysis of specific situations (case method)
9. Simulation tasks
10. Problem statement
11. Presentations
12. Trainings
13. Business games

**Control methods** – oral control, written control, test control, programmed control, practical examination, self-control, self-assessment.

**Methodical support**

1. Plans of practical classes and independent work of applicants for higher education.
2. Methodical instructions for independent work of applicants for higher education in preparation for practical training and in class.
3. Methodical materials that provide independent work for higher education.
4. Multimedia presentations.
5. Packages of clinical tasks, sets with results of paraclinical researches.
6. Test and control tasks for practical classes.
7. Questions and tasks to control the assimilation of content modules.
8. Materials for the final modular control (2 oral questions, 2 clinical tasks and evaluation criteria).
9. Demonstration materials, instructions for the use of technical teaching aids (equipment for mastering theoretical material, educational films, videos).
10. Methodical recommendations for teachers for practical classes according to the thematic plan.

It has been used the standardized generalized criteria of knowledge estimation of higher education seekers in PSMU (table 1).

**Table 1**

**Standardized generalized criteria of knowledge assessing of higher education seekers in PSMU**

| According to 4 point scale | According to ECTS | Evaluation criteria |
|---|---|---|
| 5 (excellent) | A | Higher education seekers shows special creative abilities, is able to acquire knowledge independently, without the help of the teacher finds and processes the necessary information, is |

| | | |
|---|---|---|
| | | able to use the acquired knowledge and skills for decision-making in unusual situations, convincingly argues answers, independently reveals own talents and inclinations, possesses not less than 90 % of knowledge on the topic both during the questioning and all types of control. |
| 4 (good) | B | Higher education seekers is fully oriented in the studied material, uses it in practice, freely solves exercises and problems in standardized situations, independently corrects errors, the number errors is insignificant, has at least 85% knowledge of the topic during the questioning, and all types of control . |
| | C | Higher education seekers is able to compare, summarize, systematize information under the guidance of a scientific and pedagogical worker, in general, independently use it in practice, control their own activities; to correct mistakes, among which there are significant ones, to choose arguments to confirm opinions, has at least 75% of knowledge on the topic both during the questioning and all types of control. |
| 3 (satisfactory) | D | Higher education seekers reproduces a significant part of theoretical material, shows knowledge and understanding of the basic principals with the help of a teacher can analyze educational material, correct errors, among which there are a considerable number of significant, has at least 65% knowledge of the topic, and during the questioning, and all types of control. |
| | E | Higher education seekers studied the educational material at a level higher than the initial, a significant part of it reproduces at the reproductive level, has at least 60% knowledge of the topic both during the questioning and all types of control. |
| 2 (unsatisfactory) | FX | Higher education seekers studied the material at the level of separate fragments that present a small part of the material, has less than 60% knowledge of the topic both during the questioning and all types of control. |
| | F | Higher education seekers studied the material at the level of elementary recognition and reproduction of separate facts, elements, has less than 60% knowledge of the topic as during the questionong, and all types of control. |

After completing the module study, the evaluation on traditional 4-point scale is converted into a multi-point (maximum 120 points) – the conversion of the total mark of current educational activity per module – is performed only after the class before final modular control. The conversion is performed according to the following algorithm:

– calculates the average grade of the applicant on the traditional 4-point scale, obtained during the classes of this module (to the nearest hundredth point);

– to obtain a convertible multi-point total grade of the current educational activity per module, the average grade that was obtained on the traditional 4-point scale should be multiplied by a coefficient of 24, or according to Table 2. Exceptions are cases where the average grade on the traditional 4-point scale is 2 points. In this case, the higher education seeker has 0 points on a multi-point scale;

– the average grade of the current educational activity is calculated on the total number of classes in the module, but not on the actual number of attended classes.

**Table 2**

**Unified table of correspondence of scores for current educational activity, scores for FMC, exam, and traditional four-point scale**

| Average mark for current educational activity (A) | Points for current educational activity on module (A * 24) | Points for FMC (A*16) | Points for module and / or exam (A*24 + A*16) | Category of ECTS | On 4 point scale |
|---|---|---|---|---|---|
| 2 | 48 | 32 | 80 | **F** | **2** |
| 2,1 | 50 | 34 | 84 | **FX** | **Unsatisfactory** |
| 2,15 | 52 | 34 | 86 | | |
| 2,2 | 53 | 35 | 88 | | |
| 2,25 | 54 | 36 | 90 | | |
| 2,3 | 55 | 37 | 92 | | |
| 2,35 | 56 | 38 | 94 | | |
| 2,4 | 58 | 38 | 96 | | |
| 2,45 | 59 | 39 | 98 | | |
| 2,5 | 60 | 40 | 100 | | |
| 2,55 | 61 | 41 | 102 | | |
| 2,6 | 62 | 42 | 104 | | |
| 2,65 | 64 | 42 | 106 | | |
| 2,7 | 65 | 43 | 108 | | |
| 2,75 | 66 | 44 | 110 | | |
| 2,8 | 67 | 45 | 112 | | |
| 2,85 | 68 | 46 | 114 | | |
| 2,9 | 70 | 46 | 116 | | |
| 2,95 | 71 | 47 | 118 | | |
| 3 | 72 | 50 | 122 | **E** | **3** |
| 3,05 | 73 | 50 | 123 | | **Satisfactory** |
| 3,1 | 74 | 50 | 124 | | |
| 3,15 | 76 | 50 | 126 | | |
| 3,2 | 77 | 51 | 128 | | |
| 3,25 | 78 | 52 | 130 | **D** | |
| 3,3 | 79 | 53 | 132 | | |
| 3,35 | 80 | 54 | 134 | | |
| 3,4 | 82 | 54 | 136 | | |
| 3,45 | 83 | 55 | 138 | | |

| | | | | | |
|---|---|---|---|---|---|
| 3,5 | 84 | 56 | 140 | | |
| 3,55 | 85 | 57 | 142 | | |
| 3,6 | 86 | 58 | 144 | **C** | **4 Good** |
| 3,65 | 88 | 58 | 146 | | |
| 3,7 | 89 | 59 | 148 | | |
| 3,75 | 90 | 60 | 150 | | |
| 3,8 | 91 | 61 | 152 | | |
| 3,85 | 92 | 62 | 154 | | |
| 3,9 | 94 | 62 | 156 | | |
| 3,95 | 95 | 63 | 158 | | |
| 4 | 96 | 64 | 160 | **B** | |
| 4,05 | 97 | 65 | 162 | | |
| 4,1 | 98 | 66 | 164 | | |
| 4,15 | 100 | 66 | 166 | | |
| 4,2 | 101 | 67 | 168 | | |
| 4,25 | 102 | 68 | 170 | | |
| 4,3 | 103 | 69 | 172 | | |
| 4,35 | 104 | 70 | 174 | | |
| 4,4 | 106 | 70 | 176 | | |
| 4,45 | 107 | 71 | 178 | | |
| 4,5 | 108 | 72 | 180 | **A** | **5 Excellent** |
| 4,55 | 109 | 73 | 182 | | |
| 4,6 | 110 | 74 | 184 | | |
| 4,65 | 112 | 74 | 186 | | |
| 4,7 | 113 | 75 | 188 | | |
| 4,75 | 114 | 76 | 190 | | |
| 4,8 | 115 | 77 | 192 | | |
| 4,85 | 116 | 78 | 194 | | |
| 4,9 | 118 | 78 | 196 | | |
| 4,95 | 119 | 79 | 198 | | |
| 5 | 120 | 80 | 200 | | |

The minimum convertible sum of points of current educational activity for all modules of all disciplines of all departments is uniform and is **72 points**.

**Final modular control on module** is performed upon completion of the study of all topics of the module at the last control class of the module.

Higher education seekers who have completed all types of work provided in the curriculum and scored not less than the minimum score (3.0 - 72 points) are admitted to the final control.

Higher education seekers who during the study of the module with final control, had an average score of current educational activity from 4.50 to 5.0 are exempt from FMC and automatically (by agreement) receive a final mark according to the table, however the presence of higher education seeker at FMC is mandatory. In case of disagreement with the assessment, this category of higher education seekers is performed FMC according to the general rules.

The final modular control is performed in a standardized way at the last class of the module and includes control of theoretical and practical training. Question cards for the final modular control include 2 questions (25 points each-50 points), and 2 tasks (15 points - 30 points). Each examination card question is evaluated from 0 to 25 points, practical skills quistions from 0 to 15 points.

The FMC score is evaluated in points and is not converted into a traditional 4-point score. The maximum number of FMC points is 80 points. The minimum number of FMC points at which the control is considered completed is 50 points. The maximum number of points per module is 200 points (of which up to 120 points for current educational activity).

The result of final modular control is evaluated in points (traditional 4-point evaluation is not given). The maximum number of points of final modular control is 80 points. The minimum number of points of final modular control, for which the control is considered to be passed, is 50 points. Examination cards for final modular control include 2 questions (25 points each-50 points), and 2 questions on practical skills (15 points - 30 points). Each examination card question is evaluated from 0 to 25 points, practical skills question from 0 to 15 points.

After the passing final modular control, the total number of points per module is calculated:

a) the sum of points of current educational activity;

b) scores of the final modular control.

**The maximum number of points per module is 200 points.**

Information about FMC is filled in accordance with the regulations of organization of the educational process in PSMU and submitted to the dean's office.

Applicants for higher education who have not passed the FMC have the right to retake the module twice according to the reassignment schedule.

**Recommended literature.**

**Basic (that are present in PSMU library)**

1. Medical Biology: textbook / Bazhora Yu.I., Bulyk R.Ye., Chesnokova M.M. [et al]. – Vinnytsia: Nova Knyha, 2019. – 448 p.

**Auxillary**

1. Benjamin D. Solomon MD. Medical Genetics and Genomics: Questions for Board Review. 2022. – 176 p. DOI:10.1002/9781119847212

2. Emery's. Elements of Medical Genetics, P.D. Turnpenny, S. Ellard. – Elsevier – 2017. – 400 p.

3. Hilton S. Read, M.D., Kathryn S. Read. Color Atlas of Genetics. Fourth edition, revised and updated. 2013. – 475 p.

4. Szalai, Csaba & László, Valéria & Tóth, Sára & Pap, Erna & Falus, Andras & Oberfrank, Ferenc. Medical genetics and genomics. – 2020. https://www.researchgate.net/publication/338606132_Medical_genetics_and_genomics_2019

5. Saudubray, Jean-Marie, Baumgartner, Matthias, Walter, John (Eds.) Inborn Metabolic Diseases.- Springer-Verlag Berlin Heidelberg. – 2016. – p.658.

**Information resources**

https://www.pdmu.edu.ua/n-process/department-npr/normativni-dokumenti
https://ndiseases.pdmu.edu.ua/
https://biblumsa.blogspot.com/p/blog-page_6470.html
https://bio.libretexts.org/Bookshelves/Genetics/Online_Open_Genetics_(Nickle_and_Barrette-Ng)
https://accessmedicine.mhmedical.com/book.aspx?bookID=2247
http://www.ncbi.nlm.nih.gov/omim
https://onlinelibrary.wiley.com/doi/book/10.1002/9781119847212
https://www.genome.gov/About-Genomics/Introduction-to-Genomics
https://www.testing.com/genetic-testing-techniques/

**Internet resources according to international protocols** www.nice.org.uk

**Medical literature in electronic libraries**

PubMed Embase Scirus
Google Scholar

**Open electronic medical libraries (full-text versions of articles)**

PubMed Central BioMed Central
Directory of open access journals - Health Sciences Public Library of Science - Medicine FreeMedicalJournals.com

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