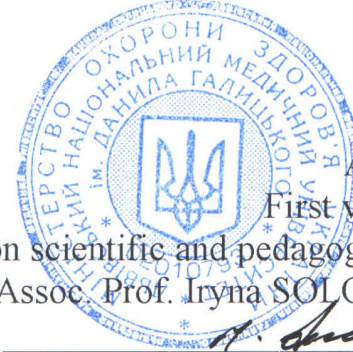


DANYLO HALYTSKYI LVIV NATIONAL MEDICAL UNIVERSITY

Department of Medical Biology, Parasitology and Genetics



Approved
First vice-rector
on scientific and pedagogical work
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“28” 06 2023

WORKING PROGRAM OF THE DISCIPLINE

«MEDICAL GENETICS»

(elective course)

ББ 1.26

for the training of specialists of the 2nd (master's degree) level of higher education
education sector 22 Public Healthcare
Specialty 221 «Dentistry»

Discussed and approved
at the methodical meeting of the
department of Medical Biology,
Parasitology and Genetics
Protocol No19 from “15” 05. 2023
Head of Medical Biology,
Parasitology and Genetics department

 Prof. Zinoviy VOROBETS

«Approved»

the profile methodical commission on
medical biological disciplines
Protocol No3
from “25” 05. 2023
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INTRODUCTION

Program of study discipline «Medical genetics»

Composed according to the higher education academic standard of the second (Master's) level

education sector 22 «Healthcare»

specialty 221 «Dentistry»

educational program of *Master* of dentistry

Brief review of the subject «Medical genetics» (abstract)

The discipline «Medical genetics» (elective course) is studied by students of the first year of study. The course is based on previously studied by students at secondary school subjects such as «General Biology», is integrated with the discipline «Medical Biology». The rapid development of medical genetics has been made possible by the development of embryology, human anatomy and physiology, cytology, biochemistry and classical genetics. The realization of the international project «Human Genome» has led to the fact that today man is one of the best studied objects of molecular genetics. In a short time, genetic diagnosis and gene therapy of many hereditary anomalies, which until recently were considered incurable, became possible. This determines the relevance of in-depth study of general genetics and medical genetics in particular.

Knowledge of the basics of medical genetics are necessary to understand the basic principle that any human pathology is to some extent related to heredity. The discipline provides general biological training for the study of modern problems and achievements of genetics, including molecular genetic diagnostics, pharmacology, gene therapy. Teaching the discipline includes lectures, practical classes, independent work of students and ends with a credit. «Medical genetics» (elective course) lays the foundation for further mastering by students of knowledge and skills in specialized theoretical and clinical professional and practical disciplines (bioorganic chemistry, pharmacology, physiology, medical genetics, clinical immunology, infectious diseases, pediatrics, etc.).

Types of classes according to the curriculum are:

a) lectures; b) practical classes, c) individual work of students; d) consultations.

Structure of the discipline	Number of credits, hours				Year of study, semester	Forms of the control
	Total	Auditory		ISW		
		Lectures	Practical classes			
Name of the discipline: «Medical genetics» Content chapters 1	3,5 credits / 105 hours	10	30	65	I course (I semester)	Credit

The subject of study of the discipline is the biological basis of human life at the molecular genetic level.

Interdisciplinary connections: discipline «Medical genetics» (elective course) is integrated with following disciplines: medical biology, biological and bioorganic chemistry, genetics, microbiology, organic and inorganic chemistry, pathological physiology, oncology, neurocybernetics, bioinformatics etc.

1. The purpose and objectives of the academic discipline.

1.1. The purpose of teaching the discipline «Medical Genetics» follows from the objectives of the educational and professional training program for graduates of higher medical education and is determined by the content of those systemic knowledge and skills that must be mastered by a dental student. The study of modern problems of molecular biology generates in students a holistic idea of the formation of knowledge and practical skills for further study by students of a block of disciplines that provide scientific and professional training for mastering modern problems and achievements of molecular medicine.

1.2. The ultimate goals of educational discipline «Medical genetics» are:

1. To explain regularities of the vital functions of organism of a human at the molecular-genetic and cellular levels.
2. To determine the displays of action of general biological laws in ontogenesis of human.
3. To understand the molecular-genetic basis for the development of hereditary and multifactorial diseases, prospects for the application of the achievements of medical genetics in practical medicine.
4. Be able to explain the nature and mechanisms of manifestation in the phenotype of hereditary human diseases.

1.3 Competencies and learning outcomes.

The discipline «Medical genetics» provides students with the acquisition of **competencies**. **Integral competence** makes it possible to apply acquired general and professional competences to solve complex tasks of a doctor's professional activity and practical problems in the field of health care in the relevant position, the scope of which is provided by defined lists of syndromes and symptoms of diseases, physiological conditions and diseases that require special tactics of patients' management; laboratory research, implementation of innovations.

- *general (3K)*:

- 3K 1. Ability to abstract thinking, analysis and synthesis.
- 3K 2. Knowledge and understanding of the subject area and understanding of professional activity.
- 3K 3. The ability to apply the acquired knowledge in their practice.
- 3K 4. Ability to communicate in the state language both orally and in writing.
- 3K 5. Ability to communicate in English.
- 3K 6. Skills in the use of information and communication technologies.
- 3K 7. Ability to search, process and analyze information from different sources.
- 3K 8. Ability to adapt and act in a new situation.

3K 9. Ability to identify, pose and solve problems.

3K 10. The ability to be critical and self-critical.

3K 11. Ability to work in a team.

3K 12. The desire to protect the environment.

3K 13. The ability to act socially responsibly and consciously.

3K 14. The ability to exercise their rights and responsibilities as a member of society, to realize the values of civil (free democratic) society and the need for its sustainable development, the rule of law, human and civil rights and freedoms in Ukraine.

3K 15. The ability to preserve and multiply moral, cultural, scientific values and achievements of society based on understanding 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.

2) *Special (ΦK):*

ΦK 2. The ability to interpret the results of laboratory and instrumental research.

ΦK 13. Ability to assess the impact of the environment on the health of the population (individual, family, population).

Details competencies according to the NQF (National Qualifications Framework) descriptors in the form of «Matrix of competencies»

№	Competence	Knowledge	Ability	Communication	Autonomy and responsibility
1	2	3	4	5	6
Integral competence					
The ability to interpret the general biological patterns that underlie the processes of human life					
Special competencies					
1.	Ability to use in the dentist's practice the knowledge of the molecular basis of heredity, the mechanisms of development of hereditary and acquired human	Molecular mechanisms of storage and realization of hereditary information. Molecular mechanisms of intercellular signaling and transmembrane transport. Molecular mechanisms of human variability	Determine the primary structure of the protein, the number of amino acids, the molecular mass of the polypeptide by the nucleotide sequence of the gene. Determine the	Be able to explain to the patient and his family the main causes of mutations and their connection with hereditary diseases. and mechanisms of tumor development	Be responsible for mastering the relevant knowledge and skills.

	diseases.	<p>Types of mutational variability.</p> <p>Molecular mechanisms of action of certain mutagenic factors and methods of studying mutagenic activity, mechanisms of action of antimutagens.</p> <p>Organization of structural eukaryotic genes, principles of regulation of gene expression in pro- and eukaryotes.</p> <p>Peculiarities of the organization of genomes of viruses, prokaryotes, eukaryotes.</p> <p>Modern methods of studying the human genome.</p> <p>Regulation of the mitotic cycle, molecular mechanisms of oncogenesis, main mechanisms of apoptosis.</p>	type of gene, chromosomal and genomic mutations.		
3.	Ability to apply knowledge of modern advances in medical genetics in practical medicine and dentistry.	<p>Modern methods of molecular genetic diagnostics and their use in medicine</p> <p>The concept of biotechnology and genetic engineering</p> <p>Principles of creation of transgenic organisms and possibilities of their use</p> <p>Principles of animal</p>	Analyze the electrophoregram of DNA and determine the presence of DNA of infectious agents, mutations in human genes.	Be able to explain to the patient and his family the essence of the methods used in molecular genetic diagnostics, the principles of obtaining recombinant drugs, the	Be responsible for mastering the relevant knowledge and skills.

		cloning and the importance of the method for biology and medicine. Principles of gene therapy, its achievements and prospects.		possibility of gene therapy of hereditary and non-hereditary diseases.	
4.	. Ability to use own professional activities to protect the environment.	Potential environmental consequences of the use of genetically modified organisms Mutagenic and antimutagenic environmental factors.	To form the requirements of environment protection.	Explain various aspects of the use of transgenic organisms, the impact of mutagenic factors on the human body, the role of man as an environmental factor; promote measures to preserve and protect the environment.	Be responsible for mastering the relevant knowledge and skills; to manifest responsible attitude and care for the environment.

Learning outcomes (ІІPH):

Integral learning outcomes of the discipline «Medical Genetics» provide an opportunity to apply the acquired general and professional competencies to solve complex problems of professional activity of doctors and practical problems in health care of the relevant position, the scope of which is provided by certain lists of syndromes and symptoms diseases that require special tactics of patient management; laboratory research, innovations.

The discipline «Medical Genetics» lays the foundation for the formation of program learning outcomes in accordance with the Standard of Higher Education of Ukraine for undergraduate training of specialists of the second (master's) level of the specialty «Dentistry»:

ІІPH 7. To analyze the epidemiological situation and carry out mass and individual, general and local drug and non-drug prevention measures for dental diseases.

ІІPH 15. To assess the impact of the environment on the state of the population health in the conditions of a medical institution according to standard methods.

ІІPH 17. To lead a healthy lifestyle, use self-regulation and self-control techniques.

ПРП 18. To be aware of and be guided in one's activities by civil rights, freedoms and duties, to raise the general educational cultural level.

Learning outcomes for the discipline

Upon completion of the discipline «Medical Genetics» students should **know**:

- Modern methods of studying the human genome.
- Classification of mutational variability, molecular mechanisms of mutational variability.
- Molecular mechanisms of action of certain mutagenic factors and methods of studying mutagenic activity, mechanisms of action of antimutagens.
- Methods of studying hereditary human diseases and diseases with hereditary predisposition.
- Classification of hereditary diseases.
- Principles of prenatal diagnosis of hereditary diseases.
- Modern methods of molecular genetic diagnostics and their use in medicine. ПОНЯТТЯ ПРО БІОТЕХНОЛОГІЮ ТА ГЕННУ ІНЖЕНЕРІЮ.
- Principles of creation of transgenic organisms, possibilities of their use in biotechnology and medicine.
- Potential environmental consequences of the use of genetically modified organisms.
- Principles of animal cloning and the importance of the method for biology and medicine.
- Principles of gene therapy, its achievements and prospects.

be able:

- to determine the types of gene mutations when solving of situational problems and in diagrams, types of chromosomal and genomic mutations when analyzing of karyotypes;
- to analyze the electrophoregram of DNA and determine the presence of DNA infectious agent, mutations in human genes;
- to predict the genotypes and phenotypes of offspring by parental genotypes;
- to calculate the probability of birth to a sick child with monogenic diseases with known genotypes of parents;
- to calculate the probability of hereditary diseases in offspring depending on the penetrance of the gene;
- to analyze the human karyotype and determine the diagnosis of the most common chromosomal diseases;
- to build a pedigree and conduct its genealogical analysis;
- to calculate the role of heredity and environmental conditions in the development of traits (based on the results of twin analysis);
- to calculate the frequencies of genes and genotypes according to Hardy-Weinberg law (based on the results of population-species method).

2. Information volume of the discipline

3,5 ECTS credits / 105 hours are allocated for the study of the discipline.

3. The structure of the discipline:

Names of content modules and topics	Hours			
	Total	Including		
		Lectures	Practical classes	IWS
Topic 1. The subject and tasks of medical genetics. The role of heredity in human pathology	18	2	4	12
Tema 2. Topic 2. Methods of human inheritance investigation.	27	2	8	17
Tema 3. Topic 3. Hereditary diseases: classification, mechanisms of occurrence.	30	2	8	20
Tema 4. Peculiarities of the human mitochondrial genome.	8	2	2	4
Tema 5. Medical and genetic counseling and prenatal diagnosis.	22	2	8	12
Total 105 hours / 3,5 credits of ECTS	105	10	30	65
Final control	Credit			

4. Topics of lectures

№	Topic	Hours
1	Subject and tasks of medical and molecular genetics.	2
2	Methods of human inheritance investigation.	2
3	Hereditary diseases: classification, mechanisms of occurrence	2
4	Peculiarities of the human mitochondrial genome.	2
5	Medical and genetic counseling and prenatal diagnostics.	2
	Total	10

5. Topics of practical classes

№	Topic	Hours
1	The subject and tasks of medical genetics. Cytological basics of human heredity.	2
2	Mutations as the main etiological factor in the development of hereditary pathology.	2
3	Drug mutagenesis, teratogenesis, carcinogenesis Testing of substances for mutagenicity.	2
4	Gene (molecular) diseases, mechanisms of their occurrence and	2

	principles of laboratory diagnostics.	
5	Characteristics of molecular diseases.	2
6	Methods of human heredity investigation: biochemical methods and their application in the study of genetic (molecular) human diseases.	2
7	Human chromosomal diseases.	2
8	General characteristics of human mitochondrial pathology. Clinic, diagnosis, treatment.	2
9	Methods of human hereditary diseases investigation	2
10	Genetics of ontogenesis	2
11	General characteristics of multifactorial diseases. Determination of genetic predisposition. Prevention measures. Basics of environmental genetics, pharmacogenetics.	2
12	Congenital defects of human development: classification, etiology, diagnosis and prevention	2
13	Methods of prenatal diagnosis of a human	2
14	Concept of gene therapy	2
15	Levels and ways of prevention of hereditary diseases	2
	Total	30

6. Individual extracurricular work

№	Topic	Hours
1	Mechanisms of genotypic variability	4
2	Mobile genetic elements. Molecular mechanisms of general genetic recombination	4
3	Characteristics of the mitochondrial genome	4
4	Immunogenetics and its practical use in medicine	4
5	Embryonic stem cells as a promising therapeutic direction of treatment	4
6	Premature aging syndromes	4
7	Modern molecular cytogenetic methods: FISH method, comparative genomic hybridization, spectral karyotyping, etc.	4
8	Recombinant DNA methods, nucleic acids hybridization.	5
9	Genome structure and general characteristics of human genes.	4
10	Mutagens Stages of mutagenesis.	4
11	Multifactorial diseases. Basics of environmental genetics.	4
12	Oncogenetics. Diagnosis of hereditary predisposition to cancer.	4
13	Disorders of amino acid metabolism: phenylketonuria, homocystinuria, albinism and alkaptonuria. Types of inheritance, clinical signs and diagnosis.	4
14	Hereditary disorders of lipid metabolism. Diseases of Tay-Sachs, Niemann-Pick, Gaucher. Causes, clinical signs and diagnosis.	4
15	Hereditary disorders of carbohydrate metabolism: galactosemia and glycogenosis. Pathogenesis and diagnosis.	4

16	Methods of prevention of hereditary diseases.	4
	Total	65

7. Individual tasks. preparation of scientific reports for the meeting of the scientific circle and for the student scientific conference.

8. Teaching methods

- Verbal methods: lecture, conversation;
- Visual methods: illustration, demonstration;
- Practical methods: performing practical work and solving situational tasks to develop skills and abilities;
- Independent work of students on comprehension and assimilation of new material;
- Use of control and educational computer programs in the discipline;

9. Methods of control

- Current control is based on the control of theoretical knowledge, practical skills and abilities at practical classes.
- The study of the discipline ends with a credit.

Criteria for evaluation. During the evaluation of the mastering of each topic for the current educational activity of the student grades are set on a 4-point (traditional) scale, taking into account the approved evaluation criteria:

- **grade "excellent" (5)** – the student has mastered the theoretical material of the topic, demonstrates deep and comprehensive knowledge of the topic, the basic principles of scientific sources and recommended literature, thinks logically and builds answers, freely uses acquired theoretical knowledge in analyzing practical material, expresses his attitude to certain problems, demonstrates a high level of practical skills;
- **grade "good" (4)** – the student has mastered the theoretical material of the class, knows the basic aspects of primary sources and recommended literature, can explain it; has practical skills, expresses his views on certain issues, but assumes certain inaccuracies and errors in the logic of the presentation of theoretical material or when performing of practical skills;
- **grade "satisfactory" (3)** – the student has mainly mastered the theoretical knowledge of the subject, is guided by primary sources and recommended literature, but unconvincingly answers, confuses concepts, additional questions cause the student uncertainty or lack of stable knowledge; answering the questions of a practical nature, reveals inaccuracies in knowledge, can not assess facts and phenomena and relate them to future activities, makes the mistakes in the implementation of practical skills;

- grade **"unsatisfactory" (2)** – the student has not mastered the study material, does not know scientific facts, definitions, almost does not navigate in primary sources and recommended literature, no scientific thinking, practical skills are not formed. The scores from discipline are converted into points.

10. Current control is carried out on the basis of a comprehensive assessment of student activities and acquired competencies (knowledge, skills, abilities, etc.), which includes control of the basic knowledge, quality of practical work, level of theoretical preparation and results of basic knowledge level control.

Current control is performed during the classes and is aimed at checking the mastering by students the learning material. The forms of current control are:

- a) tests with a choice of one correct answer (multiple choice questions), the definition of the correct sequence of actions, definition of correspondency, the definition of specific areas in a picture or diagram («recognition»);
- b) individual oral examination, interview;
- c) solving of typical situational problems;
- d) solving of typical problems from medical genetics.

10.1. Evaluation of students' current study is carried out at each practical lesson on a 4-point scale using approved criteria for evaluation of the discipline and is entered into the academic journal. All types of work and the list of competencies provided by the curriculum and methods for the study of the topic are taken into account. The student should receive a grade from each topic.

10.2. The control of the results of the tasks of independent work is carried out during the current control of the topic in the relevant lesson.

11. Final control

This is a form of final control, where assessing the student's mastery of educational material on the basis of the results of his performance of certain types of work in practical classes occurs.

Credit for discipline is conducted after the end of its study, before the examination session. All topics for current control should be submitted. Students receive a credit if the average score for current study during the semester is at least "3" (120 points on a 200-point scale).

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 students' learning activities.»

12. Scheme of accrual and distribution of points received by students

Maximum quantity of points which student for the current progress can collect during the semester makes 200 points.

Minimum quantity of points which the student should collect for current educational activity for admission to the examination makes 120 points.

The calculation of the number of points is made on the basis of the collected student's marks on the 4-points national scale during the discipline study, by calculating the arithmetic mean (AM or average), rounded to two decimal places. The obtained value is converted into points according to the scoring scale as follows:

$$x = \frac{AM \times 200}{5}$$

The scores from discipline are converted to the ECTS scale and to the 4- point scale independently. The ECTS scale points are not converted to the 4-point scale and vice versa.

Ranking with assignments of grades "A", "B", "C", "D", "E" is carried out for students of this course who study in one specialty and have successfully completed the study of the discipline.

Points of discipline for students who have successfully completed the program are converted into traditional 4-point scale by absolute criteria, which are listed in the following table:

Points from discipline	Estimation on 4-point scale
From 170 to 200 points	5
From 140 to 169 points	4
From 139 points to minimal	3
Less than minimal quantity of points, which student must collect	2

The objectivity of the assessment of students' learning activities is checked by statistical methods (correlation coefficient between ECTS assessment and assessment on a national scale).

13. Methodical support

- Curriculum of the discipline;
- Plans of lectures, practical classes and independent work of students;
- Abstracts of lectures on the discipline;
- Methodical recommendations for the teacher;
- Methodical recommendations for practical classes for students;
- Methodical materials that provide independent work of students;
- Test and control tasks for practical classes;
- Questions and tasks to control the mastering of the discipline;
- Tasks to test practical skills.

14. List of recommended literature

Basic:

1. Paryzhak S.Ya., Vorobets Z.D. Medical Biology. Textbook – Lviv: Qvart, 2020. – 426 p.

2. Bazhora Y.I., Bulyk R.Ye., Chesnokova M.M., Shevelenkova A.V., Smetyuk O.O., Lomakina Yu.V. Medical Biology: textbook. – Vinnytsia: Nova Knyha, 2018. – 448 p.: il.
3. Step 1. Lecture notes 2018 Biochemistry and Medical genetics. New York. Kaplan, Inc. – 2018 – 403 c.

Additional:

1. Bogitsh B.J., Carter C.E., Oeltmann T.N. Human parasitology. – 5th ed. Textbook. – Academic Press, 2019. – 407 p.
2. Elsheikha H.M., Jarroll E.L. Illustrated Dictionary of Parasitology in the Postgenomic Era. – Caister Academic Press, 2017. – 332 p.
3. Ghosh S., Chander J. Paniker's Textbook of Medical Parasitology. 8th Edition. – Jaypee Brothers Medical Pub, 2018. – 276 p.
4. Kaplan Medical's USMLE STEP 1. Biochemistry and Medical Genetics. Lecture notes. – 2018. – 432 p.
5. Pap E., Falus A., László V., Oberfrank F., Szalai C., Tóth S. Medical Genetics and Genomics. Edited by Typotex Kiadó. – Budapest University of Technology and Economics, 2016. – 206 p.
6. Ryabokon E.V., Onishchenko T.E., Ushenina L.O., Furyk E.A., Mashko O.P. Manual of helminthiasis: for the students of medical faculty. – Zaporozhye: [ZSMU], 2013. – 66 p.

Informational resources:

1. Testing center – database of license tests Krok – 1 <http://testcentr.org.ua/>
2. OMIM (Online Mendelian Inheritance in Man) – An Online Catalog of Human Genes and Genetic Disorders <http://omim.org/>