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
**WORKING PROGRAM OF THE DISCIPLINE**


**«GENETICS»**

(elective course)

**ББ 1.29**

**for the training of specialists of the 2nd (master's degree) level of higher education  
education sector 22 Public Healthcare  
Specialty 222 «Medicine»**

Discussed and approved  
at the methodical meeting of the  
department of Medical Biology,  
Parasitology and Genetics  
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«Approved»  
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medical biological disciplines  
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## INTRODUCTION

### Program of study discipline «Genetics»

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

education sector 22 «Healthcare»

specialty 222 «Medicine»

educational program of *Master* of medicine

#### Brief review of the subject «Genetics» (abstract)

The discipline «Genetics» (elective course) is a component of the educational-professional program, studied by students of speciality 222 «Medicine» of the first year of study.

The course is based on previously studied by students in 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 test. «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 |          |                   | IS W | Year of study, semester | Forms of the control |
|--|--------------------------|----------|-------------------|------|-------------------------|----------------------|
|  | Total                    | Auditory |                   |      |                         |                      |
|  |                          | Lectures | Practical classes |      |                         |                      |
| Name of the discipline: «Genetics»<br>Content chapters 1 | 3 credits / 90 hours     | 12       | 18                | 60   | 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:

The discipline «Genetics» (elective course) is integrated with following disciplines:

| <b>Content modules of the discipline «Genetics» (elective course)</b>                   | Disciplines with which it is integrated   |
|---|---|
| Content chapter 1. Molecular basis of heredity. Molecular basis of hereditary diseases. | <b>1. Medical biology:</b><br>a) Basis of molecular biology<br>b) Basis of molecular genetics<br><b>2. Biological and bioorganic chemistry.</b><br>Fundamentals of molecular genetics<br><b>3. Pathological physiology.</b><br>General nosology - a general study of the disease, etiology and pathogenesis. Typical pathological processes. The role of heredity, constitution, age-related changes in pathology.<br><b>4. Medical genetics.</b><br>Heredity and pathology. The role of heredity in human pathology. Monogenic diseases. Diseases with hereditary predisposition.<br><b>5 Oncology.</b> General oncology. Organization of oncological care. Epidemiology and prevention of malignant tumors. Methods of diagnosis and principles of treatment of malignant tumors. Patterns of malignant tumors development. |

### **1. The purpose and objectives of the academic discipline.**

1.1. Ultimate goals of educational discipline «Genetics» (elective course) is the formation of knowledge and practical skills for further study by students disciplines which provide natural- scientific and professionally practical preparation to master modern problems and achievements of molecular medicine.

#### 1.2. **The ultimate goals** of educational discipline are:

- To explain regularities of the vital functions of organism of a man at the molecular-genetic and cellular levels;
- To determine the displays of action of general biological laws in ontogenesis of man;

- To understand the molecular-genetic basis for the development of hereditary and multifactorial diseases and prospects for the application of the achievements of molecular biology in practical medicine.

**1.3 Competencies and learning outcomes**, formation of which promotes a discipline «Genetics» (according to the requirements of the standards of higher education).

**General (3K):**

3K 1. Ability to abstract thinking, analysis and synthesis.

3K 2. Ability to learn and acquire modern knowledge

3K 3. The ability to apply the acquired knowledge in their practice.

3K 4. Knowledge and understanding of the subject. Understanding of the professional activity.

3K 5. Ability to adapt and act in a new situation.

3K 6. Ability to make informed decisions.

3K 7. Ability to work in a team.

3K 8. Interpersonal skills interaction.

3K 9. Ability to communicate in the state language both orally and in writing.

3K 10. Skills in the use of information and communication technologies.

3K 11. Ability to search, process and analyze information from various sources.

3K 12. Definiteness and perseverance in terms of tasks and responsibilities.

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).

ΦK 17. Ability to assess the impact of the environment, social, economical and biological determinants on the health of human, children, families and populations.

ΦK 21. Communicate one's own knowledge, conclusions and arguments on health care problems and related issues to specialists and non-specialists, in particular to people who are studying, clearly and unambiguously.

ΦK 24. Adherence to ethical principles when working with patients and laboratory animals.

ΦK 25. Adherence to professional and academic integrity, to be responsible for the reliability of the obtained scientific results.

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   |
| <b>Integral competence</b>   |   |  |   |   |   |
| The ability to interpret the general biological patterns that underlie the processes of human life |   |  |   |   |   |
| <b>Special competencies</b>  |   |  |   |   |   |
| 1.   | Ability to use in the practice of the doctor's knowledge of the molecular basis of heredity, the mechanisms of development of hereditary and acquired human diseases. | Molecular mechanisms of storage and realization of hereditary information. Molecular mechanisms of intercellular signaling and transmembrane transport. Molecular mechanisms of human variability. Types of mutational variability. Molecular mechanisms of action of certain mutagenic factors and methods of studying mutagenic activity, mechanisms of action of antimutagens. Organization of structural eukaryotic genes, principles of | 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 type of gene, chromosomal and genomic mutations. | 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. |

|   |   |  |  |   |   |
|---|---|--|--|---|---|
|   |   | <p>regulation of gene expression in pro- and eukaryotes. Peculiarities of the organization of genomes of viruses, prokaryotes, eukaryotes. Modern methods of studying the human genome. Regulation of the mitotic cycle, molecular mechanisms of oncogenesis, main mechanisms of apoptosis. Molecular mechanisms of ontogenesis.</p> |  |   |   |
| 2 | <p>The ability to apply the knowledge of the peculiarities of human ontogenesis in the diagnosis and treatment of human diseases.</p> | <p>Molecular mechanisms of fertilization. Molecular mechanisms of egg activation during fertilization. The concept of totipotency, pluri- and multipotency, stem cells. Molecular bases of cell differentiation, histo- and</p>  |  | <p>Be able to explain to the patient and his family molecular-biological bases of stem cell use, biological bases of aging.</p> | <p>Be responsible for mastering the relevant knowledge and skills</p> |

|    |  |  |   |  |  |
|----|--|--|---|--|--|
|    |  | organogenesis.<br>Molecular<br>basis of aging.   |   |  |  |
| 3. | Ability to apply knowledge of modern achievements of molecular biology in practical medicine | Modern methods of molecular genetic diagnostics and their use in medicine<br>The concept of biotechnology and genetic engineering<br>Principles of creation of transgenic organisms and possibilities of their use<br>Principles of animal cloning and the importance of the method for biology and medicine.<br>Principles of gene therapy, its achievements and prospects. | 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 possibility of gene therapy of hereditary and non-hereditary diseases. | Be responsible for mastering the relevant knowledge and skills   |
| 4. | Ability to use own professional activities to protect the environment.                       | Potential environmental consequences of the use of genetically modified organisms<br>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;   | Be responsible for mastering the relevant knowledge and skills; to manifest responsible attitude and care for the environment. |



|  |  |  |  |   |  |
|--|--|--|--|---|--|
|  |  |  |  | promote measures to preserve and protect the environment. |  |
|--|--|--|--|---|--|

### Learning results:

Program learning outcomes (IPPH), the formation of which is facilitated by the discipline «Genetics» (elective course):

IPPH 1. Have thorough knowledge of 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.

IPPH 2. Understanding and knowledge of basic and clinical biomedical sciences, at a level sufficient for solving professional tasks in the field of health care.

IPPH 19. Plan and implement a system of anti-epidemic and preventive measures regarding the occurrence and spread of diseases among the population.

IPPH 21. Search for the necessary information in the professional literature and databases of other sources, analyze, evaluate and apply this information.

IPPH 23. To assess the impact of the environment on human health in order to assess the morbidity of the population.

IPPH 25. To communicate one's own knowledge, conclusions and arguments on health care problems and related issues to specialists and non-specialists in a clear and unambiguous manner.

IPPH 27. communicate fluently in the national and English languages, both orally and in writing to discuss professional activities, research and projects.

Upon completion of the discipline «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 hereditary diseases investigation 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.
- The concept of biotechnology and genetic engineering.
- 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 ECTS credits / 90 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 | 12    | 2         | 2                 | 8   |
| Topic 2. Methods of human inheritance investigation.  | 44    | 2         | 2                 | 40  |
| Topic 3. Hereditary diseases: classification, mechanisms of occurrence.                     | 18    | 4         | 10                | 4   |

|  |               |           |           |           |
|--|---------------|-----------|-----------|-----------|
| Tema 4. Peculiarities of the human mitochondrial genome.       | 8             | 2         | 2         | 4         |
| Tema 5. Medical and genetic counseling and prenatal diagnosis. | 8             | 2         | 2         | 4         |
| <b>Total 90hours / 3 credits of ECTS</b>                       | <b>90</b>     | <b>12</b> | <b>18</b> | <b>60</b> |
| <b>Final control</b>   | <b>Credit</b> |           |           |           |

#### 4. Topics of lectures

| № | Topic   | Hours     |
|---|---|-----------|
| 1 | Subject and tasks of medical and molecular genetics.                                | 2         |
| 2 | Mutations as the main etiological factor in the development of hereditary pathology | 2         |
| 3 | Peculiarities of the human mitochondrial genome.                                    | 2         |
| 4 | Hereditary diseases: classification, mechanisms of occurrence.                      | 2         |
| 5 | Methods of human inheritance investigation.   | 2         |
| 6 | Medical and genetic counseling and prenatal diagnosis.                              | 2         |
|   | <b>Total</b>  | <b>12</b> |

#### 5. Topics of practical classes

| №  | Topic  | Hours     |
|----|--|-----------|
| 1. | General characteristics of monogenic pathology. Genetics of some forms of monogenic diseases.  | 2         |
| 2. | Amino acid metabolism disorders: phenylketonuria, homocystinuria, albinism and alkaptonuria. Types of inheritance, clinical signs and diagnosis.                         | 2         |
| 3. | Hereditary disorders of carbohydrate metabolism: galactosemia and glycogenosis. Pathogenesis and diagnosis.  | 2         |
| 4. | Hereditary disorders of lipid metabolism. Diseases of Tay-Sachs, Neiman-Pick, Gaucher. Causes, clinical signs and diagnosis.   | 2         |
| 5. | General characteristics of human mitochondrial pathology. Clinic, diagnosis, treatment.  | 2         |
| 6. | Mutagens. Stages of mutagenesis. Drug mutagenesis, teratogenesis, carcinogenesis. Mutagenicity testing of substances.  |           |
| 7. | General characteristics of multifactorial diseases. Determination of genetic predisposition. Prevention measures. Fundamentals of ecological genetics, pharmacogenetics. | 2         |
| 8. | Recombinant DNA methods, nucleic acids hybridization.  | 2         |
| 9. | Levels and ways of prevention of hereditary diseases. Medical and genetic counseling and prenatal diagnosis.   | 2         |
|    | <b>Total</b>   | <b>18</b> |

## 6. Individual work

| №<br>з/п | Topic   | Hours     |
|----------|---|-----------|
| 1        | Mobile genetic elements. Molecular mechanisms of general genetic recombination                              | 4         |
| 2        | Extranuclear heredity. Mitochondrial genome   | 4         |
| 3        | Embryonic stem cells as a perspective therapeutic direction of treatment                                    | 4         |
| 4        | Genetic engineering and its methods   | 4         |
| 5        | Transgenic organisms. The principle of construction of transgenic organisms                                 | 4         |
| 6        | Transgenic bacteria, plants and animals. The main areas of application in the national economy and medicine | 4         |
| 7        | Gene therapy in oncology  | 4         |
| 8        | Nucleic acid-based vaccines and their use in medicine   | 4         |
| 9        | Genome structure and general characteristics of human genes   | 4         |
| 10       | Oncogenetics. Diagnosis of hereditary predisposition to cancer  | 4         |
| 11       | Cellular engineering. Cloning of organisms and cells  | 4         |
| 12       | Achievements of biotechnology in medicine   | 4         |
| 13       | Congenital defects: classification, etiology, diagnosis and prevention                                      | 4         |
| 14       | Nanomedicine and implementation of its achievements in medical practice                                     | 4         |
| 15       | Levels and methods of prevention of hereditary diseases   | 4         |
|          | <b>Total</b>  | <b>60</b> |

**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 test.**

**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: http://omim.org/](http://omim.org/)