

**Federal State Autonomous Educational Institution of Higher Education  
PEOPLES' FRIENDSHIP UNIVERSITY OF RUSSIA  
(RUDN UNIVERSITY)  
Institute of Medicine**

Recommended by MCSD

**ACADEMIC COURSE WORKING PROGRAM**

**Discipline: Human hereditary diseases**

**Recommended for postgraduates  
Direction 06.06.01 «Biological sciences»,  
profile «Genetics: molecular basis of human hereditary diseases»  
Qualification (degree) of a degree carrier:  
Researcher. Lecturer-researcher**

### 1. Goal and tasks of the discipline:

- to train highly qualified academic personnel;
- to form and develop their competencies in the field of medical genetics in accordance with professional standards;
- to form skills required to use the modern biological techniques and procedures

### 2. Position of the discipline in the structure of the Educational program:

Block 1 – Educational disciplines (modules). Variative part.

Preceding and following disciplines forming competencies of the discipline are shown in table 1.

Table 1. Preceding and following disciplines forming the given competencies

| №                                 | Code of competence | Preceding disciplines   | Following disciplines |
|-----------------------------------|--------------------|---|-----------------------|
| Universal competencies            |                    |   |                       |
| 1                                 | UC-1               | History and philosophy of science<br>Methodology of scientific research<br>General Genetics |                       |
| General professional competencies |                    |   |                       |
| 1                                 | GPC-1              | General Genetics  |                       |
| Professional competencies         |                    |   |                       |
| 1                                 | PC-1               | General Genetics  |                       |
| 2                                 | PC-2               | General Genetics  |                       |
| 3                                 | PC-3               |   |                       |

### 3. Requirements for the results of the discipline study

Discipline studying is designed to form the following competencies:

#### Universal competencies (UC)

- **UC-1.** the ability to analyze and evaluate current scientific achievements, generate new ideas and solve the research and practical problems.

#### GPC – general professional competencies.

- **GPC-1.** the ability to carry out research in the professional field with the use of modern methods of research and information technologies.

#### PC – professional competencies:

- **PC-1.** the ability to understand modern problems of biology and use fundamental biological ideas in professional activities for the goal setting and solution of new problems;
- **PC-2.** the ability to use the basic theories, concepts and principles in the chosen field of activity, the ability to systems thinking;
- **PC-3.** the readiness for the independent analysis of available information, the goal setting, and problem solving.

**In consequence of studying the discipline a postgraduate is due to:**

***Know:***

1. Methods used in modern genetics.
2. The concept of genetic, biochemical and structural unity of all life. The achievements of modern genetics which is one of the basic disciplines of modern biology.
3. The gene pool, the genetic unity, and genetic heterogeneity of natural populations. Genetic equilibrium in populations.
4. Chromosomes, the concept of karyotype. Morphological types of human chromosomes. Autosomes and sex chromosomes.
5. Concepts of the gene, the genotype, the genome, the phenotype. The allelic and non-allelic, linked and non-linked genes. Pleiotropic and lethal genes. The concepts of penetrance and expressivity. Forms of gene interaction.
6. The life cycle, mitotic and meiotic cell division. The control of the cell cycle.
7. The history of genetics.
8. Genotype as a system of interacting genes. The laws of heredity. Patterns of inheritance.
9. Nucleic acids as the genetic material.
10. Genetic engineering and cloning.
11. Forms of variability. Classification of mutations. Molecular mechanisms of gene mutations. The causes of mutations. Mutagens. The basic mechanisms of DNA repair.
12. The subject and tasks of Human Genetics. Man as an object of genetic research. Monogenic, polygenic, and multifactorial traits in humans.
13. Methods of Human Genetics.
14. Human hereditary diseases and their causes. Principles of diagnosis, treatment and prevention of hereditary diseases. Genetic counseling.
15. Genetic monitoring of populations. International and national programs.

***Be able to:***

1. use the methods of modern genetics, taking into account their capabilities and limitations;
2. determine the morphological types of chromosomes; write down karyotype formulas;
3. analyze the results of crosses and solve genetic problems;

4. classify mutations; analyze the causes of point mutations;
5. analyze the human karyotype in health and chromosomal diseases;
6. classify hereditary diseases;
7. evaluate the risk of pathological trait in the offspring;
8. use academic knowledge to solve different genetic problems;
9. carry out the statistical processing of the data obtained in the research;
10. write scientific work at the level required to be published in peer-reviewed journals;
11. analyze study books and scientific articles.

***Have the skills in:***

1. modern genetic techniques and procedures;
2. solving of genetic problems;
3. statistical processing of the data;
4. analysis, summarizing and public presentation of the results of the performed research work.

#### **4. Discipline hours and types of training activity**

General labor intensity is 4 points of credit

| №         | Type of academic load                                    | Totally hours | Semesters |           |
|-----------|--|---------------|-----------|-----------|
|           |  |               | 3         | 4         |
| <b>1.</b> | <b>Class exercises (academic hours)</b>                  | <b>80</b>     | <b>40</b> | <b>40</b> |
|           | Including:   |               |           |           |
| 1.1.      | Lectures   | 40            | 20        | 20        |
| 1.2.      | Practical classes  | 40            | 20        | 20        |
| <b>2.</b> | <b>Self-study work of postgraduates (academic hours)</b> | <b>64</b>     | <b>32</b> | <b>32</b> |
| <b>3.</b> | <b>Total labor intensity (academic hours )</b>           | <b>144</b>    | <b>72</b> | <b>72</b> |
|           | <i>Total labor intensity (points of credit)</i>          | 4             | 2         | 2         |

#### **5. Content of the discipline**

##### **5.1. Content of the discipline units**

| №  | Discipline unit                            | Content of the unit  |
|----|--|--|
| 1. | Introduction to Human Genetics             | Subject and tasks of Medical Genetics. Genetics in Biology and Medicine. The history of Medical Genetics.                                    |
| 2. | The object and methods of Medical Genetics | Man as an object of genetic research. Cytological basis of heredity. Methods of Human Genetics. Classification of human hereditary diseases. |

|    |   |  |
|----|---|--|
| 3. | Single gene diseases  | Genetic diseases and their classification. Mutations as basis of hereditary diseases. Most common single gene diseases   |
| 4. | Chromosomal diseases  | Chromosomes, the concept of karyotype. Morphological types of human chromosomes. Chromosomal diseases, their classification and causes. Mosaicism. Most common chromosomal diseases. |
| 5. | Multifactorial diseases                                     | Multifactorial diseases and their causes.  |
| 6. | Non-Mendelian inheritance                                   | Cytoplasmic inheritance; Genomic imprinting; Trinucleotide repeat disorders.   |
| 7. | Diagnosis, treatment and prevention of hereditary diseases. | Principles of diagnosis, treatment and prevention of hereditary diseases. Genetic counseling.  |

## 5.2. Units of the discipline and types of classes

| №  | Unit   | Lectures  | Practical classes |           | Self-study work | Totally    |
|----|--|-----------|-------------------|-----------|-----------------|------------|
|    |  |           | S                 | PC        |                 |            |
| 1. | Introduction to Human Genetics                             | 4         |                   |           | 2               | 6          |
| 2. | The object and methods of Medical Genetics                 | 10        |                   | 12        | 15              | 37         |
| 3. | Single gene diseases                                       | 6         |                   | 6         | 15              | 27         |
| 4. | Chromosomal diseases                                       | 6         |                   | 4         | 6               | 16         |
| 5. | Multifactorial diseases                                    | 2         |                   | 2         | 6               | 10         |
| 6. | Non-Mendelian inheritance                                  | 4         |                   | 6         | 10              | 20         |
| 7. | Diagnosis, treatment and prevention of hereditary diseases | 8         |                   | 10        | 10              | 28         |
|    | <b>TOTALLY:</b>  | <b>40</b> |                   | <b>40</b> | <b>64</b>       | <b>144</b> |

**6. Laboratory classes** are not provided.

## 7. Practical classes

| №  | Units                                      | Practical classes  | Labor intensity (hours) |
|----|--|--|-------------------------|
| 1. | Introduction to Human Genetics             | -  | -                       |
| 2. | The object and methods of Medical Genetics | Pedigree analysis<br>Karyotype analysis<br>Population study<br>PCR and its application<br>Sanger sequencing of DNA | 12                      |

|    |  |  |    |
|----|--|--|----|
| 3. | Single gene diseases                                       | Dominantly inherited disorders<br>Recessively inherited disorders<br>Sex-linked disorders                            | 6  |
| 4. | Chromosomal diseases                                       | Chromosome diseases  | 4  |
| 5. | Multifactorial diseases                                    | Multifactorial diseases  | 2  |
| 6. | Non-Mendelian inheritance                                  | Mitochondrial diseases<br>Nucleotide repeat expansion disorders<br>Epigenetic diseases                               | 6  |
| 7. | Diagnosis, treatment and prevention of hereditary diseases | Prenatal diagnosis of hereditary diseases<br>Prevention of hereditary diseases<br>Genetic counseling<br>Gene therapy | 10 |

## 8. Academic support:

### a) main literature

1. Concepts of genetics / W.S. Klug; Pearson new international edition. - 3th ed. - England : Pearson, 2014. - 885 p.
2. Lissauer T. Illustrated Textbook of Paediatrics / T. Lissauer, G. Clayden. - Fourth Edition - London : Elsevier, 2012. - 533 p.

### b) additional literature

1. Celehrter T.D., Collins F.S. Principles of medical genetics. – Williams and Wilkins. – 1996.
2. Gardner A., Davies T. Human Genetics. – Scion Publishing Ltd. – 2009.
3. Fletcher H., Hickey I. Genetics. – Garland Science. – 2013.
4. Lewin B. Genes. – Oxford University Press. – 2012.
5. Vogel and Motulsky's Human Genetics: Problems and Approaches / M. Speicher, Antonarakis S.E., Motulsky A.G. – Springer. – 2010.

## 9. Data bases, information, reference and search systems:

1. <http://esystem.rudn.ru/>
2. National Center for Biotechnology Information (NCBI) - [www.ncbi.nlm.nih.gov](http://www.ncbi.nlm.nih.gov)
3. ScienceDirect - <http://www.sciencedirect.com>
4. Scientific electronic library: - <http://elibrary.ru>
5. Google Academy - <http://scholar.google.ru/>

## 10. Equipment and material support of the discipline:

- Computers
- Multimedia projectors
- The PCR laboratory

## **11. Methodological recommendations on discipline study organization:**

During practical classes and lectures corresponding topics are studied with the use of computers and multimedia projectors.

Before each class PhD students must read the corresponding topic in the recommended main and additional text-books and try to answer questions for self-study control.

Self-study work is organized in class rooms of the department and in the computer class.

Electronic study guides on some topics are also posted in the Internet on the site of the department:

<http://esystem.pfur.ru/>

Out-of-class self-study work includes:

- Learning topics with the use of text-books, study guides;
- Preparation of reports on the selected topic
- Preparation for tests

## **12. Evaluation instrument fund**

Evaluation instrument fund for the discipline «Human hereditary diseases» is available for students at TUIS RUDN.

The program is designed in accordance with the RUDN educational standard of higher education.

**Developers of the training and methodology complex**



M.M. Azova



O.B. Gigani

**Director of the program**



M.M. Azova