

**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: Methods in Human Genetics

**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 molecular and 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 Methodology of scientific research 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 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. Man as an object of genetic research.
6. Methods of Human Genetics (pedigree analysis, twin study, karyotyping, biochemical techniques, population study, DNA analysis).

Be able to:

1. use the methods of modern genetics, taking into account their capabilities and limitations;
2. determine the morphological types of chromosomes;
3. analyze the results of crosses and solve genetic problems;
4. carry out the polymerase chain reaction, and analyzing the results of the experiment in different ways;
5. analyze the human karyotype;
6. use methods of Human Genetics to analyze genetic material and patterns of inheritance;
7. evaluate the risk of pathological trait in the offspring;
8. solve 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
1.	Class exercises (academic hours)	80	40	40
	Including:			
1.1.	Lectures	40	20	20
1.2.	Practical classes	40	20	20
2.	Self-study work of postgraduates (academic hours)	64	32	32
3.	Total labor intensity (academic hours)	144	72	72
	<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 Human Genetics. Genetics in Medicine. Man as an object of genetic research.
2.	Pedigree analysis	Tasks of the method. Drawing up a family tree using the standard symbols. Patterns of inheritance. Solving of genetic problems.
3.	Twin study	Tasks and importance of the method. Multifactorial diseases. Solving of genetic problems.
4.	Karyotype analysis	Techniques or karyotyping and their tasks. Preparing karyotypes from mitotic cells. Banding techniques. Karyotype formula in health and diseases. Fluorescence in situ hybridization (FISH).

5.	DNA analysis	Tasks of different methods. DNA and RNA extraction. The polymerase chain reaction. Gel electrophoresis. Types of the PCR. Restriction enzymes and their application. DNA sequencing. Southern and Northern blotting.
6.	Biochemical techniques	Principles of biochemical diagnosing of human hereditary diseases.
7.	Population study	Tasks of the method. Genetic equilibrium in populations. The Hardy–Weinberg Law. Solving of genetic problems.

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.	Pedigree analysis	8		8	12	28
3.	Twin study	2		2	2	6
4.	Karyotype analysis	6		6	20	32
5.	DNA analysis	12		20	20	52
6.	Biochemical techniques	4		2	6	12
7.	Population study	4		2	2	8
TOTALLY:		40		40	64	144

6. Laboratory classes are not provided.

7. Practical classes

№	Units	Practical classes	Labor intensity (hours)
1.	Introduction to Human Genetics	-	-
2.	Pedigree analysis	Drawing up a family tree using the standard symbols. Patterns of inheritance.	8
3.	Twin study	Twin study and its application	2
4.	Karyotype analysis	Banding techniques. Fluorescence in situ hybridization (FISH) Comparative genomic hybridization (CGH)	6
5.	DNA analysis	The polymerase chain reaction Primer design	20

		Real time PCR Gel electrophoresis Restriction enzymes and their application. Sanger sequencing. Next generation sequencing (NGS) Genome-editing technique Methods for analysis of DNA methylation	
6.	Biochemical techniques	Biochemical techniques in Human Genetics	2
7.	Population study	Analysis of the genetic structure of a population	2

8. Academic support:

a) main literature

1. Concepts of genetics / W.S. Klug; Pearson new international edition. - 3th ed. - England : Pearson, 2014. - 885 p.

b) additional literature

1. Gardner A., Davies T. Human Genetics. – Scion Publishing Ltd. – 2009.
2. Fletcher H., Hickey I. Genetics. – Garland Science. – 2013.
3. Lewin B. Genes. – Oxford University Press. – 2012.
4. Vogel and Motulsky's Human Genetics: Problems and Approaches / M. Speicher, Antonarakis S.E., Motulsky A.G. – Springer. – 2010.
5. Color Atlas of Genetics / Passarge Eberhard. - 4th edition, revised and update . - Stuttgart ; New York : Thieme, 2013.
6. Genetics / Friedman, M, D., Fred J., Hayden, M.B., Michael R. - Baltimore : Harwal Publishing Company, 1992. - 250 p. : ill. - (The National Medical Series for Independent Study).

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
3. Nature Genetics - <http://www.nature.com/ng/index.html>
4. Royal Society of Chemistry - <http://pubs.rsc.org/>
5. ScienceDirect - <http://www.sciencedirect.com>
6. Scientific electronic library: - <http://elibrary.ru>
7. 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 «Methods in Human Genetics» 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