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INTERNATIONAL EUROPEAN
UNIVERSITY



SCHOOL OF
MEDICINE

Modern methods of genetic diagnosis

2021



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Subject matter 			
		Modern methods of genetic diagnosis	
Lecturer (s) 			
		Professor of the Department of Fundamental Disciplines Oksana Voitovska	
Lecturer's profile 			
		https://medicine.ieu.edu.ua/pro-yemsh/kafedry/kafedra-fundamentalnykh-dystsyplin	
Consultations			
On-campus consultations	The second Wednesday of the month from 14:00 to 15:00		
On-line consultations		The third Tuesday of the month from 17:00 to 18:00	
Contact phone number 			
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Discipline page 			
			
Form of final control	Final test	Grading test	Exam
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>



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1 Subject matter summary

According to the curriculum, the study of the elective discipline "Modern methods of genetic diagnostics" is provided in third year in the fifth semester.

According to modern medicine, any human pathology is more or less related to heredity. This provision is the basis for teaching and studying medical genetics as a clinical and preventive discipline. Because heredity and variability are integral parts of life, genetics should be the basis of theoretical and clinical training of physicians.

The need for genetic knowledge for the doctor is also determined by the constant increase in the proportion of hereditary pathology in the structure of morbidity, mortality and disability of the population. Elective course "Modern methods of genetic diagnosis" introduces students to modern basic methods of diagnosis of hereditary pathology.

2 Background of studying the subject matter

The study of the elective course "Modern methods of genetic diagnosis" is provided in the third year in the fifth semester, when students acquire relevant knowledge in basic fundamental disciplines: medical biology, medical and biological physics, human anatomy and physiology, bioorganic and biological chemistry, and began studying microbiology, virology and immunology, with which the curriculum is integrated.

3 Aim and objectives of the disciplines

The aim for teaching the discipline "Modern methods of genetic diagnosis" is to lay the foundations for students to study modern diagnostic technologies used not only in the diagnosis of hereditary diseases, but also in general clinical practice, which integrates teaching with different disciplines and skills to apply knowledge of modern methods, genetic diagnostics in the process of further training and professional activity; to establish an understanding of modern features of monogenic and chromosomal diseases, as well as common human diseases that arise against the background of hereditary predisposition and require the integration of classical clinical concepts and modern high technology.

Objectives:

- to provide students with knowledge about modern diagnostic methods used in medical genetics.
- to teach students to solve typical and complex specialized problems and practical problems in professional activities in the field of health care, or in the learning process, which involves research and / or innovation and is characterized by complexity and uncertainty of conditions and requirements.

4 Study results

As a result of studying the discipline "Modern methods of genetic diagnosis" the student must know:

- risk groups for the development of hereditary diseases;
- algorithm of examination of patients of high genetic risk for the development of hereditary diseases;
- indications for invasive prenatal diagnosis.

As a result of studying the discipline "Modern methods of genetic diagnostics" the student must be able to:

- identify risk groups for the development of hereditary diseases;
- to determine the algorithm of examination of patients of high genetic risk for the development of hereditary diseases;
- analyze and interpret the results of the survey; analyze the condition of the fetus;



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- to determine the indications for invasive prenatal diagnosis

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ECTS credits

The study of the discipline is given 92 hours (10 hours of lectures, 36 practical classes, 50 hours of VTS), 3 ECTS credits. The discipline is studied in the third year in the fifth semester.



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6 Subject matter structure			
Name of topics	Type of classes / year		
	Lectur	Practical /Seminar	Individual work
<i>SECTION I. SYNDROMOLOGICAL ANALYSIS. MODERN CYTOGENETIC METHODS OF DIAGNOSIS OF CONGENITAL AND HEREDIC PATHOLOGY.</i>			
<i>Semantic section 1. Syndromological analysis</i>			
Topic 1. Methodology of examination of a patient with suspected hereditary pathology. Analysis of phenotypic features of the proband and his family members	-	-	2
Topic 2. Clinical and genealogical analysis. Methods of compiling a pedigree	1	2	2
Topic 3. Syndromological analysis. Application of syndromological analysis in the diagnosis of hereditary pathology	1	2	2
<i>Semantic section 2. Cytogenetic methods of diagnosis of congenital and hereditary pathology</i>			
Topic 4. Structure and functions of chromosomes.	-	-	2
Topic 5. Cytogenetic research methods in the clinic. Chromosomal abnormalities (numerical, structural).	2	2	2
Topic 6. Chromosomal polymorphism, chromosomal instability gonadal mosaicism, single-parent disomy..	-	2	2
Topic 7. Molecular cytogenetic methods of diagnosis (FISH)	-	1	2
Topic 8 .DNA sequencing	-	1	2
<i>SECTION II. MODERN BIOCHEMICAL METHODS OF DIAGNOSIS OF CONGENITAL AND HEREDIC PATHOLOGY</i>			
<i>Content section 3. Biochemical methods of diagnosis of congenital and hereditary pathology.</i>			
Topic 9. Diagnosis of hereditary metabolic diseases substances.	2	2	2



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6 Subject matter structure			
Name of topics	Type of classes / year		
	Lectur	Practical /Seminar	Individual work
Topic 10. Mass screening programs in the early diagnosis of hereditary pathology.	-	2	3
Topic 11. Selective screening programs in the diagnosis of SHO.	-	2	2
Topic 12. Modern methods of clarifying the diagnosis of SHO. Interpretation of high performance liquid chromatography results		2	2
Topic 13. Modern methods of clarifying the diagnosis of SHO. Interpretation of gas chromatography - mass spectrometry results	-	1	2
Topic 14. Modern methods of clarifying the diagnosis of SHO. Interpretation of tandem mass spectrometry results	-	1	2
Topic 15. Interpretation of the results of enzymatic diagnosis of lysosomal storage diseases	-	-	2
SECTION III. MODERN MOLECULAR-GENETIC METHODS OF DIAGNOSIS OF HERITABLE PATHOLOGY			
Semantic section 4. Molecular genetic methods of diagnosis of hereditary pathology.			
Topic 16. Structure and functions of DNA	-	-	2
Topic 17. Modern methods of DNA diagnosis of hereditary pathology	2	2	2
Topic 18. The latest technologies in molecular diagnostics (DNA analysis on microchips)	-	2	4
Content section 5. Prenatal diagnosis of congenital and hereditary pathology.			



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Topic 19. Methods of prenatal diagnosis	2	2	4
Topic 20. Prenatal ultrasound diagnosis of congenital malformations	-	2	4
Topic 21. Invasive methods of prenatal diagnosis.	-	2	2
Topic 22. Laboratory methods of prenatal diagnosis.	-	2	2
TOTAL OF THE DISCIPLINE - 92	10	32	50

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List of obligatory tasks

1. Clinical and genealogical method.
2. Methods of compiling a pedigree.
3. Types of inheritance.
4. Mitochondrial heredity.
5. What does cytogenetics study?
6. Indications for cytogenetic analysis.
7. Define the concept of karyotype.
8. Identify types of genomic mutations.
9. Identify types of chromosomal mutations.
10. Determine the indications for cytogenetic and molecular genetic research.
11. Classification of hereditary metabolic diseases.
12. General characteristics of SHO.
13. Methods, possibilities of methods of biochemical diagnostics.
14. Biochemical methods in the early diagnosis of SHO.
15. Indications and conditions for conducting mass screening programs.



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8 List of selective tasks

1. Creating multimedia presentations on practical topics
2. Creating crossword puzzles on the topics of practical classes
3. Participation in the work of the student scientific circle
4. Participation in the student Olympiad of the discipline
5. Participation in student scientific-practical conferences
6. Organization and visiting of thematic museums
7. Publication of abstracts of scientific conference reports in co-authorship with the teacher.

9 Subject matter features

Term of teaching	Semestr	International disciplinary integration	Course (year of study)	Cycles:: general training / vocational training / free choice
I Semestr	V	Yes	3 course	Cycle of elective disciplines

10 Assessment system and requirements

The current success of students is assessed on a 4-point scale (2; 3; 4; 5) on each practical class, taking into account the approved assessment criteria for the discipline. The student must receive a grade from each topic for further conversion of grades into points on a multi-point (200-point) scale.

Criteria for evaluating current learning activities:

Excellent ("5") - the student answered 90-100% of the questions correctly. He solves situational problems of increased complexity, is able to summarize the material.

Good ("4") - the student answered 70-89% of the questions correctly. He has the necessary practical skills and techniques to perform them in excess of the required minimum.

Satisfactory ("3") - the student answered 50-69% of the questions correctly. He has only a mandatory minimum of research methods.

Unsatisfactory ("2") - the student answered 50% of the questions correctly. He makes significant, gross mistakes when answering and demonstrating practical skills.

Assessment of independent work of students in preparation for classroom practical classes is carried out during the current control of the topic in the relevant classroom.

The final control of knowledge of the discipline is carried out in the form of a test.

The test is evaluated on a two-point scale (passed / not credited) and a 200-point scale by determining the arithmetic mean of current grades for each practical lesson on a 4-point scale, and its subsequent conversion into points on a 200-point scale. The minimum number of points that an applicant must score is 120

QR Code: <https://ieu.edu.ua/docs/rate-of-study.pdf>

11 Subject matter policy

Students who have completed all types of work, tasks provided for in the curriculum for the semester in accordance with the discipline, attended all provided classes in the curriculum, written and passed a medical history and have an average score for current academic activity of at least "3 "(72 points on a 120-point scale).

<https://ieu.edu.ua/docs/rate-of-study.pdf>



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Subject matter policy

Basic principles of classes:

- openness to new and extraordinary ideas, tolerance, friendly partnership atmosphere of mutual understanding and creative development;
- all tasks provided by the program must be completed in a timely manner;
- to achieve the goals of training and successful completion of the course, it is necessary from the first day to be actively involved in work, regularly attend lectures and practical classes, prepare and teach protocols for practical classes, not be late for classes, come to class dressed in a medical gown, be in the classroom without outer clothing and hat (for necessity to cover the head, it is allowed to wear a medical cap).

The student must complete all the necessary tasks in class and work daily on self-improvement, be able to work in a team in a mini-group, seek help and receive it when you need it. Various models of work in the classroom, including work on solving problems gives the opportunity to reveal their own potential, learn to trust their partners, to develop skills of intellectual work in a team.

- the course involves intensive use of mobile learning technologies and information exchange in joint groups of mobile messengers, which allows students and teachers to communicate with each other at any time convenient for them, and for students who are absent from classes to get the necessary educational information and present completed tasks. On the other hand, students should exclude the possibility of using a mobile phone during tests and checking homework, tablet or other mobile devices, do not resort to writing and plagiarism, adhere to the cooperation and solidarity of teacher and students, ask the teacher for help and consultations on scientific, and research work, to take part in scientific circles;
- It is forbidden to come to classes with a sharp tobacco smell. If a student smokes, the odor must be eliminated and kept in a protective medical mask throughout the class;
- Throughout the course, students' autonomous skills are actively developed, which can prepare additional information on a topic that is not included in the list of topics of practical classes of content modules and make a presentation or additional information. The teacher, in turn, must ensure the full implementation of the curriculum, not to be late for lectures, practical (seminar) classes, to objectively assess the knowledge and practical skills of students. It is important to prevent any manifestations of corruption, to pay special attention to students in practical classes while working with the equipment, to prevent prejudice and discrimination regardless of race, ethnicity and religious beliefs.

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Policy of absence and late task performance

A student who, for valid reasons, was not subject to the current control of the masses, which is documentary proved, is entitled to pass the current control within two weeks after returning to study.

A student who was absent from classes without good reason, did not participate in current control activities, did not eliminate academic debt, is not allowed to the final semester assessment on the discipline, and on the day of the exam in the examination sheet the scientific and pedagogical worker writes "rejected". Re-taking the test on the discipline is appointed subject to the implementation of all types of educational, independent (individual) work provided by the working curriculum of the discipline and is carried out in accordance with the approved by the directorate schedule of liquidation of academic debt.

<https://ie.u.edu.ua/docs/050.pdf>



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14 Academic honesty policy

Participants in the educational process are guided by the principles of academic integrity
<https://ieu.edu.ua/docs/011.pdf>

15 Recommended information sources

Main literature:

1. Grechanina O.Ya., Bogatyreva RV, Bilovol OM and co-authors "Clinic and genetics of hereditary diseases accompanied by gastrointestinal and general abdominal symptoms." Ternopil, TSMU, 2008. - 216 p.

2. Kozlova SI, Semanova E., Demikova IS, Blinnikova OE Hereditary syndromes and medical and genetic counseling. Directory. - L.: Meditsina, 2012 .

Additional literature:

1. Medical genetics. Textbook for students of higher medical (pharmaceutical) educational institutions III-IV / Edited by Grechanina O.Ya., Bogatyreva RV, Volosovets OP: K., 2007 - 535 p.

2. Artamonov RG Rare diseases in pediatrics. Diagnostic algorithms. - M.: GEOTAR- Media ». - 2012. - 128 p.

3. Baranov AA, Borovik TE, Ladodo KS, Bushuyeva TV, Grechanina OY, Maslova OI, Kuzenkova LM, Chumakova OV, Studenikin VM, Dzvonkova NG, Timofeeva AG, Kon IY, Novikov PV, Zdybska OP, Grechanina Yu.B., Lebedinets IO Hereditary disorders of amino acid metabolism. Moscow-Kharkiv, 2013, 126 p.

4. Barashnev YI, Bakharev VA, Novikov PV. Diagnosis and treatment of congenital and hereditary diseases in children. - M., "Triad-X", 2009.

5. Bochkov NP. Clinical genetics. Textbook. M. Medicine, 2010.

Additional information resources:

1. Scientific Bulletins of the Institute of Molecular Biology and Genetics: <https://www.imbg.org.ua/uk/journals/>

2. Scientific Bulletin of the Ukrainian Society of Geneticists and Breeders: <http://utgis.org.ua/index.php/ua/publ-ua/visnyk-ua>

3. Bulletin of problems of biology and medicine: <https://vpbm.com.ua/ua/>

4. Georg F. Hoffmann, Johannes Zschocke. Vademecum Metabolicum, 2015.

If you want to be successful in this discipline, you need to:

16 Tips for successful learning of the course

1. Be active, persistent, inquisitive, consistent

2. Be neat and polite

3. Systematically prepare for practical classes

4. Attend lectures and keep notes

5. Perform tasks for independent work and defend them in class.

6. Carefully handle the equipment of the department, including microscopes.

7. Be present in a medical gown at the class

8. Solve tests and problems independently, actively work in class.

9. Visit the site of the Step Center and focus on issues of medical biology.

10. Prepare presentations and crossword puzzles on the subject. Participate in student scientific conferences and engage in research work in scientific circles of the department.

11. I wish you perseverance, determination and motivation to study. And then success will come to you! See you in class!