

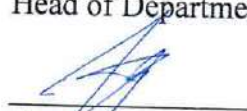
**Kharkiv National Medical University**  
**Educational and Scientific Institute of Postgraduate Education**  
**Department of Medical Genetics**  
**Medicine**  
**Educational program for training specialists of the second (master's)**  
**level of higher education training 22 "Health"**  
**in specialty 222 "Medicine"**

**2021-2022 year**

**SYLLABUS OF THE ELECTIVE COURSE**  
**MODERN METHODS OF GENETIC DIAGNOSIS**

The syllabus of the discipline was approved  
at a meeting of the Department of Medical  
Genetics  
Protocol from  
"27" August 2021 №13

Head of Department


  
\_\_\_\_\_  
(signature)

Grechanina Yu.B.  
(surname and initials)

"27" August 2021

Approved by the methodical commission of  
KhNMU on problems of professional training  
therapeutic profile  
Protocol from  
"31" August 2021 №1

Chairman of the methodical commission of  
KhNMU on problems of professional training  
therapeutic profile

  
\_\_\_\_\_  
(signature)

Kravchun P.G.  
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"31" August 2021

**Developers:** Grechanina Yuliya Borysivna

**Teachers:**

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**Інформація про викладачів:**

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**Live consultations:** by prior arrangement;

**Online consultations:** Google Meet system, Moodle system, ZOOM system according to the schedule;

**Location:** classes are held in the KNP CHO "Interregional Specialized Medical and Genetic Center - Center for Rare (Orphan) Diseases" ("MSMHC-CR (O) Z" (Independence Ave., 13), remotely - in Google Meet, MOODLE or ZOOM

**Online consultations:** schedule and venue by prior arrangement with the teacher.

**Location:** classes are held in the conditions of KNP CHO "MSMGTs-CR (O) Z" (Independence Ave., 13).

**Discipline information**

Name of indicators	Field of knowledge, direction of training, educational and qualification level	Characteristics of the discipline
		full-time education
Number of credits - 3.0	educational program for training specialists of the second (master's) level of higher education training 22 "Health"	Elective course
The total number of hours is 90	Specialty: 222 "Medicine"	<b>Year of preparation:</b> 3th
		<b>Semester</b> 5/6
Hours for day (or evening) form of study: classrooms - 20 independent work of the student - 70	Education level: master	<b>Lectures</b> 10 Hours
		<b>Practical, seminar</b> 10 Hours
		<b>Laboratory</b> 0 Hours
		<b>Individual work</b> 70 hours
		<b>Individual tasks:</b>
		Type of control:
		Credit



### Description of the discipline (abstract).

The course «MODERN METHODS OF GENETIC DIAGNOSIS» is designed for 3th year students. During the course, practical classes are held, covering a wide range of important problems of medical genetics.

The purpose of teaching the discipline «MODERN METHODS OF GENETIC DIAGNOSIS» is to acquire and deepen knowledge, skills, abilities and other competencies in medical genetics required in professional activities, which are established on the basis of educational and professional program: determining the risk group for hereditary diseases, determining the algorithm of high genetic risk for the development of hereditary diseases, analysis and interpretation of the results of cytogenetic, biochemical, molecular genetic tests.

The main objectives of the discipline "Medical Genetics" are the acquisition by students of competencies in accordance with the general and professional competencies of the educational-professional program "Medicine" of the second (master's) level of higher education in 222 Medicine qualification master of medicine: possession of survey skills, ability to determine the required laboratory list -instrumental research and evaluation of their results, establishing a preliminary and clinical diagnosis of the disease, determining the necessary mode of work and rest, the nature of nutrition in the treatment of diseases, determining the principles and nature of treatment of diseases, medical manipulations, medical records, ability to apply knowledge in practical situations, understanding of the subject area and professional activity, the ability to adapt and act in a new situation, making an informed decision, the ability to work in a team, to act socially responsible and consciously.

*Prerequisites.* The study of the discipline involves the prior mastering of disciplines in medical biology, normal and pathological anatomy, normal and pathological physiology, biochemistry, microbiology.

*Postrequisites.* The main provisions of the discipline should be applied in the study of related disciplines during the 3 years of study.

**Objective:** to provide training for highly qualified specialists in the field of medicine, namely in medical genetics, able to solve complex problems of diagnosis of congenital and hereditary pathology.

**The main objectives of the course** are the acquisition by students of competencies in accordance with the general and professional competencies of the educational-professional program "Medicine" of the second level of higher education in the specialty 222 Medicine (discipline "Medicine")

• **Integrated competencies:**

ability to solve typical and complex specialized tasks and practical problems in professional activity in the field of health care, or in the process of training, which involves research and / or innovation and is characterized by complexity and uncertainty of conditions and requirements.

• **General competencies:**

ability to abstract thinking, analysis and synthesis, ability to learn and be modernly trained; ability to apply knowledge in practical situations; knowledge and understanding of the subject area and understanding of professional activity; ability to adapt and act in a new situation; ability to make an informed decision; work in a team; interpersonal skills; ability to communicate in the state language both orally and in writing; ability to communicate in a foreign language; skills of using information and communication technologies; determination and persistence in terms of tasks and responsibilities; ability to act socially responsibly and consciously.

• **Professional competencies in the field of medical genetics:**

Survey skills; ability to determine the necessary list of laboratory and instrumental studies and



evaluate their results; ability to establish a preliminary and clinical diagnosis of the disease; ability to determine the necessary mode of work and rest, the nature of nutrition in the treatment of diseases; ability to determine the principles and nature of disease treatment; ability to diagnose emergencies; skills of performing medical manipulations; ability to determine the tactics of management of persons subject to dispensary supervision; ability to keep medical records.

The study of this discipline forms in students of social skills:

- communicativeness (implemented through: the method of group work and brainstorming during the analysis of clinical cases, the method of presenting the results of independent work and their defense in the group),
- teamwork (implemented through: group work method and brainstorming during the analysis of clinical cases),
- conflict management (implemented through: business games),
- time management (implemented through: the method of self-organization during classroom work in groups and independent work),
- leadership skills (implemented through: the method of presenting the results of independent work and their defense in the group).

**Discipline status:** elective course; the format of the discipline is mixed - the discipline, which has support in the Moodle system, teaching the discipline, provides a combination of traditional forms of classroom learning with elements of distance learning, which uses available interactive information technologies (ZOOM, Moodle, Google Meet), face-to-face and distance counseling.

**Teaching methods.**

Clinical (supervision of patients with hereditary pathology and suspicion of it), electronic information (presentations, video materials, methodical recommendations, lectures), scientific (participation in scientific developments in the discipline), control (tests, situational tasks, assessment of practical skills, defense of a clinical case).

**Learning outcomes.**

The course covers the main aspects of training a future family doctor, pediatrician, geneticist.

According to the training program in the discipline «MODERN METHODS OF GENETIC DIAGNOSIS», the applicant will acquire theoretical knowledge, methodological training, practical skills and abilities in the following areas:

Syndromological analysis.

Cytogenetic methods of diagnosis of congenital and hereditary pathology.

Biochemical methods of diagnosis of congenital and hereditary pathology.

Molecular genetic methods for the diagnosis of hereditary pathology.

Prenatal diagnosis of congenital and hereditary pathology.

**The content of the discipline  
Curriculum of the discipline**

<b>Lecture topics</b>		
1.	Methodology of examination of a patient with suspected hereditary pathology. Analysis of phenotypic features of the proband and his family members. Syndromological analysis	2
2.	Cytogenetic research methods in the clinic	2
3.	Diagnosis of hereditary metabolic diseases	2
4.	Modern technologies in molecular diagnostics	2
5.	Methods of prenatal diagnosis of congenital and hereditary pathology	2
	<b>Overall</b>	<b>10</b>



<b>Topics of practical classes</b>		
1.	Clinical and genealogical analysis. Methods of compiling a pedigree	2
2.	Chromosomal polymorphism, chromosomal instability, gonadal mosaicism, single-parent disomy	2
3.	Mass screening programs in the early diagnosis of hereditary pathology	2
4.	Modern methods of DNA diagnostics	2
5.	Diagnosis of hereditary metabolic diseases	2
	<b>Overall</b>	<b>10</b>

<b>Themes of independent works</b>		
1.	Methodology of examination of a patient with suspected hereditary pathology. Analysis of phenotypic features of the proband and his family members	2
2.	Clinical and genealogical analysis. Methods of compiling a pedigree	2
3.	Syndromological analysis. Application of syndromological analysis in the diagnosis of hereditary pathology	4
4.	Structure and functions of chromosomes	2
5.	Cytogenetic research methods in the clinic. Chromosomal abnormalities (numerical, structural)	2
6.	Chromosomal polymorphism, chromosomal instability, gonadal mosaicism, single-parent disomy	2
7.	Molecular cytogenetic diagnostic methods (FISH)	4
8.	DNA sequencing	2
9.	Diagnosis of hereditary metabolic diseases	2
10.	Mass screening programs in the early diagnosis of hereditary pathology	2
11.	Selective screening programs in the diagnosis	4
12.	Modern methods of clarifying the diagnosis of hereditary metabolic diseases. Interpretation of high performance liquid chromatography results	4
13.	Modern methods of clarifying the diagnosis of hereditary metabolic diseases. Interpretation of gas chromatography - mass spectrometry results	4
14.	Modern methods of clarifying the diagnosis of hereditary metabolic diseases. Interpretation of tandem mass spectrometry results	4
15.	Interpretation of results of diagnostics of lysosomal diseases of accumulation	4
16.	Structure and functions of DNA	4
17.	Modern methods of DNA diagnosis of hereditary pathology	2
18.	Modern technologies in molecular diagnostics (DNA analysis on microchips)	2
19.	Methods of prenatal diagnosis of congenital and hereditary pathology	2
20.	Prenatal ultrasound diagnosis of congenital malformations	2
21.	Invasive methods of prenatal diagnosis	4
22.	Laboratory methods of prenatal diagnosis	4
23.	Individual independent work of students	6
	<b>Overall</b>	<b>70</b>

### **List of questions for the differential test**

#### **1. Syndromological analysis**

1. Define the concept - an innate morphogenetic variant.
2. Define the concept of congenital malformation.
3. Identify the features of clinical manifestations of hereditary pathology.
4. To determine the algorithm of examination of the patient and his family in hereditary pathology.



5. What are the rules of genealogy?
6. Define the concept of penetrance and gene expression.
7. What are the signs of autosomal dominant inheritance?
8. What are the signs of autosomal recessive inheritance?
9. What are the signs of mitochondrial inheritance?
10. What are the signs of X-linked dominant and recessive inheritance?
11. Define the concept - syndromic analysis.

## ***2. Cytogenetic methods of diagnosis of congenital and hereditary pathology***

1. What does cytogenetics study?
2. Indications for cytogenetic analysis
3. Define the concept of karyotype.
4. Identify types of genomic mutations.
5. Identify the types of chromosomal mutations.
6. Determine the indications for cytogenetic and molecular cytogenetic studies.
7. What are the indications for determining the level of chromosomal instability?
8. Which cells can be the subject of cytogenetic studies?
9. Types of differentiated chromosome staining?
10. What are the advantages of differential staining of chromosomes?
11. What is a centromere index?
12. How to write the karyotype of the proband with increasing size of the satellites of the 22nd chromosome?
13. How to write the karyotype of a proband when doubling the satellites of the 15th chromosome?
14. How to write the karyotype of the proband with increasing length of the satellite filament of chromosome 13?
15. How to write the karyotype of a proband when doubling the satellite strand of chromosome 14?
16. How to write a karyotype of a proband at increase in length of a heterochromatin site in the 3rd chromosome?
17. How to record the karyotype of a proband in trisomy 13?
18. How to write the karyotype of a proband in monosomy X?
19. How to record the karyotype of a proband in trisomy X?
20. How to write a karyotype of a proband at duplication In?

## ***3. Biochemical methods of diagnosis of congenital and hereditary pathology***

1. Classification of hereditary metabolic diseases.
2. General characteristics of hereditary metabolic diseases.
3. Indications for screening.
4. Techniques, possibilities of methods of biochemical diagnostics. Biochemical methods in the early diagnosis of hereditary metabolic diseases.
5. Indications and conditions for conducting mass screening programs.
6. Mass screening for phenylketonuria.
7. Mass screening for congenital hypothyroidism.
8. Mass screening for adrenogenital syndrome.
9. Mass screening for cystic fibrosis.
10. Indications for selective screening programs.
11. Methods used in selective biochemical screening
12. Selective screening for hereditary amino acid metabolism diseases.
13. Selective screening for hereditary diseases of carbohydrate metabolism.
14. Selective screening for hereditary connective tissue metabolic diseases.
15. Selective screening for organic aciduria.

16. Selective screening for hereditary diseases of purine and pyrimidine metabolism.
17. Selective screening for hereditary diseases of metal metabolism.
18. Selective screening for cystic fibrosis.
19. Biochemical diagnostic methods for neuromuscular pathology.
20. Biochemical methods of diagnosis in mitochondriopathies.
21. Biochemical methods of diagnosis of disorders of fat metabolism.
22. Biochemical diagnostic methods for catastrophes of the perinatal period.
23. High-performance liquid chromatography as a method of clarifying the diagnosis of hereditary metabolic diseases.
24. Gas chromatography - mass spectrometry as a method of clarifying the diagnosis of hereditary metabolic diseases.
25. Tandem mass spectrometry as a method of clarifying the diagnosis of hereditary metabolic diseases.

#### ***4. Molecular genetic methods for the diagnosis of hereditary pathology***

1. What are the methods of DNA diagnosis of hereditary pathology?
2. What are the indications for DNA diagnostic methods?
3. What are the latest technologies in molecular diagnostics?
4. What is the structure of the mitochondrial genome?
5. What do direct and indirect methods of DNA diagnostics include?
6. What is the structure and function of DNA?
7. What is the genetic code?
8. What are the types of mutations?
9. What methods are used to detect mutations?
10. DNA analysis on microchips.

#### ***5. Prenatal diagnosis of congenital and hereditary pathology***

1. Definition of prenatal diagnosis.
2. The main tasks of prenatal diagnosis.
3. What are the methods of prenatal diagnosis?
4. What is the strategy of ultrasound examination of the fetus?
5. What are the timing of screening for birth defects?
6. What are the indications for examination at the second and third levels of prenatal diagnosis?
7. Indications, conditions and contraindications to invasive prenatal diagnosis. Які можливі ускладнення після проведення інвазивної діагностики?
8. Terms of chorionic biopsy, amniocentesis, placentocentesis, cordocentesis.
9. Marker ultrasound signs of chromosomal pathology.
10. Diagnostic value of biochemical markers of prenatal diagnosis.
11. The value of Doppler in prenatal diagnosis, indications for its implementation.



### Recommended reading

1. Medical genetics. Textbook for students of higher medical (pharmaceutical) educational institutions III-IV / Edited by Grechanina O.Ya., Bogatyreva RV, Volosovets OP: K., 2010 - 535p.
2. Metabolic diseases: a textbook / Grechanina YB, Grechanina EY, Beletskaya SV - Kharkiv: KhNMU, 2016. - 204 p.
3. Kozlova SI, Semanova E., Demikova IS, Blinnikova OE Hereditary syndromes and medical and genetic counseling. Directory. - L.: Meditsina, 2013.
4. Georg F. Hoffmann, Johannes Zschocke. Vademecum Metabolicum, 2015.
5. McKusick VA Mendelian inheritance in man. 10th ed. v.1, 2. Johris Hopkins Univ.Press. 2014. <http://www.ncbi.nlm.nih.gov/omim>

### Discipline policy and values.

In order to successfully complete the relevant course, it is necessary to regularly attend practical classes; to have theoretical preparation for practical classes according to the subject; not to be late and not to miss classes; perform all necessary tasks and work in each lesson; be able to work with a partner or in a group; contact the curators of the course on various issues on the subject of classes and receive it when you need it.

Students can discuss different tasks, but their performance is strictly individual. It is not allowed to write off, use various software, tips, use a mobile phone, tablet or other electronic gadgets during classes for purposes other than the educational process. Students are not allowed to be late for practical classes.

Curation of patients is possible provided that students have the appropriate form of clothing, a health book with a mark on vaccination against diphtheria, the results of the examination for the stress of immunity to measles (or a mark on vaccination).

Students with special needs can meet with the teacher or warn him before the start of classes, at the request of the student it can be done by the head of the group. If you have any questions, please contact the teacher.

Students' participation in research and conferences on this topic is encouraged.

All students of KhNMU are protected by the Regulations on Prevention, Prevention and Settlement of Cases Related to Sexual Harassment and Discrimination at Kharkiv National Medical University, designed to define an effective mechanism for resolving conflict situations related to discrimination and sexual harassment. This Regulation is developed on the basis of the following normative legal acts of Ukraine: the Constitution of Ukraine; Law of Ukraine "On Education"; Law of Ukraine "On Higher Education"; Law of Ukraine "On Principles of Preventing and Combating Discrimination in Ukraine"; Law of Ukraine "On Ensuring Equal Rights and Opportunities for Women and Men"; Convention for the Protection of Human Rights and Fundamental Freedoms; Convention for the Suppression of Discrimination in Education; Convention on the Elimination of All Forms of Discrimination against Women; General Recommendation № 25 to paragraph 1 of Article 4 of the Convention on the Elimination of All Forms of Discrimination against Women; General Comment № 16 (2005) "Equal rights for men and women to enjoy economic, social and cultural rights" (Article 3 of the International Covenant on Economic, Social and Cultural Rights; UN Economic, Social and Cultural Rights Committee); Recommendations on education in the spirit of international understanding, cooperation and peace and education in the spirit of respect for human rights and fundamental freedoms (UNESCO); The concept of the State social program to ensure equal rights and opportunities for women and men for the period up to 2021. Kharkiv National Medical University provides education and work that is free from discrimination, sexual harassment, intimidation or exploitation. The University recognizes the importance of confidentiality. All persons responsible for the implementation of this policy (staff of deans, faculties, institutes and



the Center for Gender Education, members of the student government and ethics committee, vice-rector for research and teaching) are confidential about those who report or accuse of discrimination, or sexual harassment (except where the law requires disclosure and / or when disclosure by the University is necessary to protect the safety of others).

KhNMU creates a space of equal opportunities free from discrimination of any national, racial or ethnic origin, sex, age, disability, religion, sexual orientation, gender, or marital status. All rights, privileges, programs and activities granted to students or staff of the University apply to all without exception, provided they are properly qualified. The anti-discrimination policy and the policy of counteracting sexual harassment of KhNMU are confirmed by the Code of Corporate Ethics and the Charter of KhNMU.

### **The behavior in the classroom**

It is important for students to follow the rules of good behavior at the university. These rules are common to all, they also apply to all faculty and staff, and are not fundamentally different from the generally accepted norms.

During classes

• allowed:

- leave the audience for a short time if necessary and with the permission of the teacher;
- drink soft drinks;
- take photos of presentation slides;
- take an active part in the class.

• prohibited:

- eat (except for persons whose special medical condition requires another - in this case, medical confirmation is required);
- smoking, drinking alcohol and even low-alcohol beverages or drugs;
- use obscene language or use words that offend the honor and dignity of colleagues and faculty;
- gambling;
- damage the material and technical base of the university (damage inventory, equipment; furniture, walls, floors, litter the premises and territories);
- shouting, shouting or listening to loud music in classrooms and even in corridors during classes.

### **Plagiarism and academic integrity**

The Department of Medical Genetics maintains zero tolerance for plagiarism. Students are expected to constantly raise their awareness of academic writing. The first lessons will provide information on what to consider plagiarism and how to properly conduct research and scientific research.

### **Occupational Health**

The first lesson of the course will explain the basic principles of labor protection by conducting appropriate training. It is expected that every higher education seeker should know where the nearest evacuation exit to the audience, where the fire extinguisher is, how to use it, etc.

**Procedure for informing about changes in the syllabus:** necessary changes in the syllabus are approved by the methodical commission of KhNMU on the problems of professional training of therapeutic profile and published on the site of KhNMU, the site of the Department of Medical Genetics of KhNMU.



## Evaluation policy

**Organization of current control.** Teachers make sure that each student receives the necessary competence in the areas included in the topics of practical classes. Assimilation of the topic (current control) is controlled in a practical lesson in accordance with specific goals. The following tools are used to assess the level of preparation of students: tests, solving situational problems, interpretation and evaluation of laboratory tests, methods of prescribing therapy, monitoring the acquisition of practical skills. Assessment of current learning activities (CPA) in each practical lesson is carried out on the traditional 4-point scale: "excellent", "good", "satisfactory" and "unsatisfactory".

**Grade from the discipline.** The final lesson is conducted in accordance with the curriculum during the semester on schedule, during classes. The grade in the discipline is given to the student at the last (final) lesson. The final score for PND and the final lesson is defined as the arithmetic mean of traditional grades for each lesson and PZ, rounded to 2 decimal places (to the nearest hundredth), which are converted into points in accordance with the "Instructions for assessing student learning. ... »Using Table 2 or the average grade (to the nearest hundredth) for HDPE and its conversion into ECTS scores, the teacher automatically receives using the electronic journal ACS. The minimum number of points that a student must score for the current activity during the study of the discipline is 120 points, the maximum number of points - 200 points.

**Assessment of students' independent work.** Independent work of students, which is provided by the topic of the lesson along with the classroom work, is assessed during the current control of the topic in the relevant lesson.

**Assessment of individual student tasks** carried out for the performance of the teacher's tasks (report of the abstract in a practical lesson, report at clinical conferences on the basis of the department, writing abstracts, articles, participation in competitions and conferences) - 1 point

During the assessment of mastering each academic topic of the discipline and the final lesson, the student is graded according to the traditional 4-point system: "excellent", "good", "satisfactory" and "unsatisfactory".

The maximum number of points that a student can score when studying the discipline is - 200, the minimum - 120 points.

After graduating from the discipline "Modern methods of genetic diagnostics" the student receives a test.

### Recalculation of the average score for current activities in a multi-point scale

1. Evaluation of current educational activities. Recalculation of the average grade for current activities in a multi-point scale is carried out in accordance with the "Instructions for assessing the educational activities of students" (Table 2).

2. Assessment of the discipline. Defined as the arithmetic mean of traditional grades for each lesson and software, rounded to 2 decimal places, which are converted into points using Table 2.



**Recalculation of the average score for current activities in a multi-point scale  
(for disciplines ending with a credit)**

4-point scale	200-point scale	4-point scale	200-point scale	4-point scale	200-point scale
5	200	4.22-4,23	169	3.45-3,46	138
4.97-4,99	199	4.19-4,21	168	3.42-3,44	137
4.95-4,96	198	4.17-4,18	167	3.4-3,41	136
4.92-4,94	197	4.14-4,16	166	3.37-3,39	135
4.9-4,91	196	4.12-4,13	165	3.35-3,36	134
4.87-4,89	195	4.09-4,11	164	3.32-3,34	133
4.85-4,86	194	4.07-4,08	163	3.3-3,31	132
4.82-4,84	193	4.04-4,06	162	3.27-3,29	131
4.8-4,81	192	4.02-4,03	161	3.25-3,26	130
4.77-4,79	191	3.99-4,01	160	3.22-3,24	129
4.75-4,76	190	3.97-3,98	159	3.2-3,21	128
4.72-4,74	189	3.94-3,96	158	3.17-3,19	127
4.7-4,71	188	3.92-3,93	157	3.15-3,16	126
4.67-4,69	187	3.89-3,91	156	3.12-3,14	125
4.65-4,66	186	3.87-3,88	155	3.1-3,11	124
4.62-4,64	185	3.84-3,86	154	3.07-3,09	123
4.6-4,61	184	3.82-3,83	153	3.05-3,06	122
4.57-4,59	183	3.79-3,81	152	3.02-3,04	121
4.54-4,56	182	3.77-3,78	151	3-3,01	120
4.52-4,53	181	3.74-3,76	150	Less than 3	Not enough
4.5-4,51	180	3.72-3,73	149		
4.47-4,49	179	3.7-3,71	148		
4.45-4,46	178	3.67-3,69	147		
4.42-4,44	177	3.65-3,66	146		
4.4-4,41	176	3.62-3,64	145		
4.37-4,39	175	3.6-3,61	144		
4.35-4,36	174	3.57-3,59	143		
4.32-4,34	173	3.55-3,56	142		
4.3-4,31	172	3.52-3,54	141		
4.27-4,29	171	3.5-3,51	140		
4.24-4,26	170	3.47-3,49	139		

Head of the department  
medical genetics,  
d.med.n., professor



Grechanina Yu.B.