

MINISTRY OF HEALTH OF UKRAINE
KHARKIV NATIONAL MEDICAL UNIVERSITY
Department of Medical Genetics
Academic year 2022-2023

SYLLABUS OF THE EDUCATIONAL COMPONENT

_____«ACTUAL ISSUES OF MEDICAL GENETICS»_____

Educational component _____ selective _____

Form of education _____ full-time education _____

Education training _____22 Health _____

Specialty _____223 «Nursing»

Educational-professional program _____«Nursing»_____

First (bachelor`s) level of higher education

Course ___2 (4 years of study)___

The syllabus of the discipline was
approved at a meeting of the
Department of Medical Genetics
Protocol from
«27» August 2021 № 1

Approved by the methodical commission
of KhNMU on problems of professional
training therapeutic profile
Protocol from
«27» August 2021 № 1

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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).

Description of the discipline (abstract).

The syllabus of the educational component "Actual issues of medical genetics" is compiled in accordance with the educational and professional program (hereinafter referred to as "Nursing Program") and the Standard of Higher Education of Ukraine (hereinafter referred to as the Standard), first (bachelor's) level, field of knowledge 22 "Health Care ", specialty "Nursing".

The course «Actual issues of medical genetics» is designed for 2th year students. During the course, practical classes are held, covering a wide range of important problems of medical genetics.

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

The general concept of teaching medical genetics in higher medical institutions is to integrate genetic knowledge and clinical thinking of the future doctor. In this regard, pedagogical activities should be aimed primarily at helping students to actively consciously use previously acquired theoretical knowledge of genetics in clinical practice, replenish knowledge of medical and clinical genetics, especially modern problems of diagnosis, treatment and prevention of hereditary pathology, the study of a number of "new" common nosological forms of hereditary diseases.

The purpose of teaching the discipline «Actual issues of medical genetics» 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 «Actual issues of medical genetics» are the acquisition by students of competencies in accordance with the general and professional competencies of the educational-professional program «Nursing» of the first (bachelor`s) level of higher education in 223 Nursing qualification bachelor 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, propaedeutics of pediatrics, pharmacology and medical recipes.

Postrequisites. The main provisions of the discipline should be applied in the study of related disciplines during the 5 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 "Nursing" of the first level of higher education in the specialty 223 Nursing (discipline "Nursing")

• **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 apply knowledge in practical situations; knowledge and understanding of the subject area and understanding of professional activity; ability to make an informed decision.

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

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.

The study of the educational component ensures that students acquire the following learning outcomes: to conduct a nursing subjective and objective examination of various organs and systems of the patient and to evaluate the obtained data. In the conditions of health care institutions and at home, by communicating with patients of different age groups, their relatives or close relatives of a sick child and his parents, be able to collect complaints, medical history, life history, collect information for compiling a genealogy, evaluate anamnestic data.

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: normative; 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 "Actual issues of medical genetics", the applicant will acquire theoretical knowledge, methodological training, practical skills and abilities in the following areas:

- Heredity and pathology. The role of heredity in human pathology.
- Methods of medical genetics.
- Propaedeutics of hereditary pathology. Methodology of phenotype description. Syndromological analysis.
- Monogenic and epigenetic diseases.
- Chromosomal diseases.
- Mitochondrial diseases.
- Diseases with hereditary predisposition.
- Prevention of hereditary pathology. Medical and genetic counseling and prenatal diagnosis.

2.1. 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 first (bachelor`s) level of higher education training 22 "Health"	Elective course
The total number of hours is 90	Specialty: 223 «Nursing»	Year of preparation:
		2th
		Semester
		3
Hours for day form of study: classrooms - 36 independent work of the student -54	Education level: Bachelor	Lectures
		10 Hours
		Practical, seminar
		26 Hours
		Laboratory
		0 Hours
		Individual work
54 hours		
		Individual tasks:
		Type of control: Credit

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

MEDICAL GENETICS COURSE (90 hours) 3.0 credit		
Topics of lection classes (10 hours)		
1.	Subject and tasks of medical genetics. The role of heredity in human pathology. Semiotics of hereditary diseases.	2
2.	General characteristics of monogenic pathology.	2
3.	General characteristics of chromosomal diseases.	2
4.	General characteristics of mitochondrial pathology.	2
5.	Levels and ways of prevention of hereditary diseases.	2
	Overall	10
Topics of practical classes (20 hours)		
1.	Subject and tasks of medical genetics. The role of heredity in human pathology.	2
2.	Clinical and genealogical method. Cytogenetic and molecular genetic methods. Biochemical methods.	3
3.	Semiotics of hereditary diseases. Features of manifestations of hereditary diseases. Morpho-genetic variants of development. Developmental defects.	2
4.	General characteristics of monogenic pathology. Clinic and genetics of some forms of monogenic and epigenetic diseases.	3
5.	Hereditary metabolic diseases. Principles of treatment of hereditary diseases, rehabilitation and social adoption.	3
6.	General characteristics of chromosomal diseases. Clinic of the main forms of chromosomal diseases.	3
7.	General characteristics of mitochondrial pathology. Clinic, diagnosis, treatment.	3
8.	General characteristics of multifactorial diseases. Determination of genetic predisposition. Prevention measures.	3
9.	Levels and ways of prevention of hereditary diseases. Medical and genetic counseling. Prenatal diagnosis. Screening programs.	2
10.	Credit	2
	Overall	26
Themes of independent works (54 hours)		
1.	Preparation for a practical lesson on the topic №1 "Subject and objectives of medical genetics. The role of heredity in human pathology.	2
2.	Preparation for a practical lesson on the topic №2 "Clinical and genealogical method. Cytogenetic and molecular genetic methods. Biochemical methods»	2
3.	Methodology of examination of a patient with suspected hereditary pathology. Analysis of phenotypic features of the proband and his family members	4
4.	Syndromological analysis. Application of syndromological analysis in the diagnosis of hereditary pathology	4
5.	Preparation for a practical lesson on the topic №5 "Hereditary metabolic diseases. Principles of treatment of hereditary diseases, rehabilitation and social adaptation "	2

6.	Modern methods of clarifying the diagnosis of hereditary metabolic diseases. Interpretation of high performance liquid chromatography results	4
7.	Modern methods of clarifying the diagnosis of hereditary metabolic diseases. Interpretation of gas chromatography - mass spectrometry results	4
8.	Interpretation of results of diagnostics of lysosomal diseases of accumulation	4
9.	Modern technologies in molecular diagnostics.	4
10.	Preparation for a practical lesson on the topic №6 "General characteristics of chromosomal diseases. Clinic of the main forms of chromosomal diseases "	2
11.	Підготовка до практичного заняття за темою №7 «Загальна характеристика мітохондріальної патології. Клініка, діагностика, лікування».	2
12.	Preparation for a practical lesson on the topic №8 "General characteristics of multifactorial diseases. Determination of genetic predisposition. Prevention measures	2
13.	Preparation for a practical lesson on the topic №9 "Levels and ways of prevention of hereditary diseases. Medical and genetic counseling. Prenatal diagnosis. Screening programs "	2
14.	Hereditary kidney disease	4
15.	Systemic skeletal dysplasia	4
16.	Fundamentals of ecological genetics, pharmacogenetics.	4
17.	Individual 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.	4
	Overall	54

Recommended reading

1. Medical genetics. Textbook for students of higher medical (pharmaceutical) educational institutions III-IV / Edited by Grechanina O.Ya., 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 2022. 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 ECTC 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		