

**MINISTRY OF HEALTH OF UKRAINE  
ODESSA NATIONAL MEDICAL UNIVERSITY**

**Department of clinical immunology, genetics and medical biology**

**SYLLABUS OF THE EDUCATIONAL DISCIPLINE  
" MEDICAL GENETICS"**

<b>The scope of the academic discipline</b>	Total number of hours per discipline: 90 hours, 3,0 ECTS credits VII - VIII semester IV year of study
<b>Days, time, place</b>	According to the approved schedule in the auditoria of clinical immunology, genetics and medical biology department, Olhivska str., 4
<b>Lecturers</b>	Shevelenkova Alla Vladimirovna: Ph.D. (Medicine), associate professor Chesnokova Marina Mikhailivna: Ph.D. (Medicine), associate professor
<b>Contact information</b>	Help by phone: Shevelenkova Alla: (097) -27-65-754 Chesnokova Marina: (048)728-54-74, (097) 256-10-33 E-mail: Shevelenkova Alla: <i>shevel2003@ukr.net</i> Chesnokova Marina: <i>marchesn2005@gmail.com</i> Face-to-face consultations: every Thursday – from 14:00 till 17:00, every Saturday – from 09:00 till 13:00. <i>For distance online studying consultations: every Thursday – from 14:00 till 17:00, every Saturday – from 09:00 till 13:00 on the Microsoft Teams, Zoom, Telegram, Viber platforms</i>

### **COMMUNICATION**

Communication with students will be carried out through face-to-face meetings.

In case of transition to distance online learning, communication with students will be carried out by phone, e-mail and *Microsoft Teams, Zoom, Telegram, Viber* platforms.

### **ANNOTATION OF EDUCATIONAL DISCIPLINE**

*The subject of study* of the educational discipline "Medical genetics" is hereditary diseases and pathology with a hereditary predisposition in humans.

*Course prerequisites and post-requisites (the place of the academic discipline in the educational program).*

*Prerequisites:* the educational discipline "Medical Genetics" is based on the study by students of the I-III courses of such disciplines as "Medical Biology", "Biological and Bioorganic Chemistry", "Pathological Physiology", "Pathomorphology", "Propaedeutics of Internal Medicine".

*Postrequisites:* the academic discipline "Medical genetics" lays the foundations for studying such disciplines as "Pediatrics", "Obstetrics and gynecology", "Forensic medicine"

*The main purpose of discipline "Medical genetics"* is the formation of knowledge and practical skills in the field of etiology, pathogenesis, clinical manifestations, diagnostics and prevention of hereditary pathology.

*The main tasks of studying the discipline* are

1. To explain the etiology of chromosomal, single gene and multifactorial diseases.
2. To recognize the general manifestations of hereditary pathology, to diagnose the congenital morphogenetic variants, to use correctly the appropriate terminology in describing the clinical picture and phenotype of the patient.
3. To select the patients for cytogenetic, specific biochemical and molecular genetic diagnosis.

4. To make a preliminary conclusion on genetic risk in the most common single gene and chromosomal diseases

5. To identify high-risk groups for the development of hereditary diseases

6. To propose preventive measures of hereditary and congenital diseases.

*Learning outcomes for discipline:*

Upon completion of the study of the discipline "Medical genetics" students must

*know:*

- classification of mutations and mutagenic factors;
- concept, effects of genomic imprinting,
- lethal effects of mutations (significance in perinatal, early childhood and infant mortality, association with infertility, spontaneous abortion),
- classification of hereditary pathology,
- morphogenetic variants and their significance in the diagnosis of hereditary syndromes and inborn states.
- etiology, pathogenesis, classification of congenital defects, classification of teratogenic factors,
- principles and stages of clinical-genealogical examination.
- characteristic of pedigrees with different types of inheritance (autosomal-dominant, autosomal-recessive, X-linked, mitochondrial)
- general symptoms of chromosomal diseases,
- peculiarities of the clinical manifestations of individual syndromes: Down, Patau, Edwards, "cat cry", Prader-Willi, Angelman, Shereshevsky-Turner, Klinefelter, polysomy X, polysomy Y.
- indications for cytogenetic and molecular-cytogenetic diagnostics,
- medical genetic counseling in case of chromosomal diseases, the potentiality of prenatal diagnosis of chromosomal diseases,
- general questions of etiology and pathogenesis of single gene diseases, classification of single gene diseases,
- a general description of single gene diseases with different types of inheritance,
- clinical characteristics, genetics, and diagnosis of Marfan syndrome, achondroplasia, congenital hypothyroidism, phenylketonuria, fragile X- syndrome, Duchen-Becker muscular dystrophy,
- general characteristics and classification of inborn errors of metabolism,
- basic methods for diagnosis of suspected inborn errors of metabolism,
- indications for molecular-genetic and biochemical diagnostics,
- principles of selection of nosological forms for screening preclinical diagnostics,
- classification and general characteristics of multifactorial disorders,
- examples of multifactorial disorders with polygenic and monogenic predisposition,
- groups of genes that participate in oncogenesis,
- definition and examples of oncogenetic syndromes (OGS),
- levels of prophylaxis of hereditary diseases, ways of prevention,
- indications for medical genetic counseling (MGC), the purpose and objectives of the MGC,
- principles of preconceptional prevention,
- methods of prenatal diagnosis, basic principles and purpose of prenatal screening,
- deontological problems of MGC, prenatal diagnosis and screening genetic programs.

*To be able:*

- to conduct a conversation with the patient for collection of the genealogic history, to compose and analyze the pedigree,
- to be able to detect minor anomalies typical for chromosomal disorders, to reveal the symptoms of the most common single gene diseases (according to list 2) during the physical examination of the patient,

- to analyze the karyograms of patients with the most common chromosomal diseases, determine the type of structural or numerical chromosomal aberration.
- to identify indications for molecular-genetic, cytogenetic and biochemical diagnostics of hereditary diseases.
- to identify indications for medical genetic counseling.
- to calculate genetic risk in the most common hereditary diseases

## DESCRIPTION OF EDUCATIONAL DISCIPLINE

### *Forms and methods of teaching*

The educational discipline "Medical genetics" will be taught in the form of practical classes (30 hours), and organization of independent work of students (60 hours).

### *Teaching methods:*

Practical lessons: heuristic conversation, explanation, discussion, case studies, practical work.

Independent work: independent work with recommended basic and additional literature, with electronic information resources, independent work with a bank of test tasks, independent solving of cases.

### Content of the academic discipline

Topic 1. Classification and etiology of hereditary diseases.

Topic 2. Semiotics of hereditary diseases.

Topic 3. General characteristics of chromosomal diseases. Cytogenetic methods

Topic 4. Chromosomal diseases caused by numerical and structural aberrations of autosomes.

Topic 5. Chromosomal diseases caused by numerical and structural aberrations of sex chromosomes.

Topic 6. General characteristics and classifications of single gene disorders. Genealogical method.

Topic 7. Single gene disorders with autosomal-dominant and X-linked modes of inheritance.

Topic 8. Single gene disorders with X-linked mode of inheritance.

Topic 9. Single gene disorders with autosomal-recessive mode of inheritance. Inborn errors of metabolism.

Topic 10. General characteristics and classification of multifactorial diseases.

Topic 11. Methods of laboratory diagnosis of hereditary disorders

Topic 12. Levels and ways of prevention of hereditary diseases.

Topic 13. Medical-genetic counseling. Credit

### *List of recommended materials*

#### *Main:*

1. Methodical recommendations on medical genetics

#### *Additional:*

1. Genetics in medicine. - 7<sup>th</sup> edition/Robert L/Nussbaum, Roderick R. McInnes, Huntington F. Willard. – 2007 – 585 p.

2. Emery's Elements of medical genetics. 15th ed. / Peter Turnpenny, Sian Ellard. – Elsevier, 2017. – 400 pp.

3. Lynn B. Jorde, John C. Carey, Michael J. Bamshad. Medical genetics. 5th ed. Elsevier, 2016. 356 pp.

4. Vogel and Motulsky's human genetics. Problems and approaches / M. R. Speicher, S. E. Antonarakis, F. G. Motulsky. 4th addition. – Springer, 2010. – 981 pp.

5. Young Ian.D. Medical genetics. -2<sup>nd</sup> ed. -Oxford university press, 2010. - 304 p.

6. Diseases of the fetus and newborn. Pathology, radiology and genetics. G.B.Reed, A.E. Claireaux and A.D.Bain., - Great Britain, - 1989, 812 p.
7. Human molecular genetics. Tom Strachan, Andrew P.Read. – 4<sup>th</sup> edition - Bios Scientific Publisher, 2010, 680 p.
8. Smith recognizable patterns of human malformation. Seventh edition. John M. Graham, - USA, - 2013, 976 p.
9. R. Wiedemann, K.-R. Gross, H.Dibberin ,Atlas of characteristic syndromes. A Visual Aid to Diagnosis. Second edition. -London, 1986,412 p.

*Information resources:*

<https://ghr.nlm.nih.gov> National library of medicine, genetics

<https://www.orpha.net> The portal for rare diseases and orphan drugs

<https://rarediseases.org> National Organization for Rare Disorders

<http://omim.org>/OMIM (Online Mendelian Inheritance in Man) – An Online Catalog of Human Genes and Genetic Disorder

## EVALUATION

*Forms and methods of current control:* current control is carried out on the basis of control of theoretical knowledge, practical skills and abilities: oral survey, testing, assessment of the performance of practical work, solving situational tasks, assessment of activity in class.

### Current assessment criteria for practical training:

«5»	The student is fluent in the material, takes an active part in the discussion and case studies, confidently demonstrates practical skills during performing and interpreting of practical work on the topic of the lesson, expresses his opinion on the topic of the lesson.
«4»	The student is well versed in the material, participates in the discussion and case studies, demonstrates practical skills while performing practical work with some mistakes, expresses his opinion on the topic of the lesson.
«3»	The student does not have enough knowledge on theoretical material, insecurely participates in the discussion and case studies, performs practical work with significant errors.
«2»	The student does not have knowledge on theoretical material, does not participate in the discussion and case studies, does not demonstrate practical skills during the practical work on the topic of the lesson.

*Forms and methods of final control:* credit is issued to the applicant who completed all sections of the educational program of the selected discipline, took an active part in practical classes, has an average current grade of at least 3.0 and has no academic debt. At the last lesson of the semester, the teacher is obliged to put "done" in the student's record book.

*Possibility and conditions of obtaining additional (bonus) points:* not provided.

### Distribution of points received by higher education applicants

The grade obtained for the answer to the exam and the score of the average current performance during the study of the discipline are used to calculate the arithmetic mean, which is the overall grade for the discipline. The average score for the discipline is transformed into a national grade and converted into scores on a multi-point scale(200-point scale).

Converting of the traditional grade from the discipline to 200-point is performed by the information and computer center of the university program "Contingent" by the formula: average grade point of success (current /in discipline) x 40

Table of conversion of traditional assessment to multi-point assessment

National grade for the discipline	Total grades for the discipline
Excellent «5»	185-200
Good «4»	151-184
Satisfactorily «3»	120-150
Unsatisfactorily «2»	Less than 120

According to the ECTS rating scale, the achievements of students in the educational

component who study in the same course of the same specialty are evaluated, according to the points they received, by ranking, namely:

Conversion of the traditional grade from the discipline and the sum of points on the ECTS scale

ECTS Point	Statistics indicator
«A»	The best 10 % of the students
«B»	Next 25 % students
«C»	Next 30 % students
«D»	Next 25 % students
«E»	Next 10 % students

## **INDEPENDENT WORK OF HIGHER EDUCATION APPLICANTS**

Independent work involves preparation for each practical lesson.

### **COURSE POLICY**

#### *Deadlines and Rework Policy:*

- Absences of classes for non-respectable reasons are worked out according to the schedule of the teacher on duty.
- Absences due to valid reasons are processed according to an individual schedule with the permission of the dean's office.
- Unsatisfactory assessment from the final practical lesson is rewritten on the days of consultations and exercises; in the case of distance learning - in the terms determined and agreed with the teacher.
- The rewriting of an unsatisfactory grade from the final test control is carried out according to a schedule agreed with the Educational and Production Complex of Innovative Technologies of Learning, Informatization and Continuous Education of the Odessa National Medical University
- Retaking the exam is carried out according to the schedule agreed with the dean.

#### *Academic Integrity Policy:*

Applicants must observe academic integrity, namely:

- independent performance of all types of work, tasks, forms of control provided for by the work program of this educational discipline;
- references to sources of information in the case of using ideas, developments, statements, information;
- compliance with the legislation on copyright and related rights;
- provision of reliable information about the results of one's own educational (scientific) activity, used research methods and sources of information.

Unacceptable in educational activities for participants of the educational process are:

- using family or official ties to obtain a positive or higher grade during any form of control of learning outcomes or academic performance;
- use of prohibited additional materials or technical means (cheat sheets, notes, micro-earphones, telephones, smartphones etc.) during control measures;
- passing procedures for control of training results by fake persons

For violation of academic integrity, students may be held to the following academic responsibility:

- a decrease in the results of assessment of control work, assessment in class, credit, etc.;
- retaking the assessment (control work, credit, etc.);
- appointment of additional control measures (additional individual tasks, control works, tests, etc.);
- conducting an additional inspection of other works authored by the violator.

*Attendance and Tardiness Policy:*

Attending all types of classroom classes (lectures, practical classes) is mandatory.

Uniform: a medical gown that completely covers the outer clothing, or medical pajamas, a cap, a mask, and a change of shoes.

Equipment: notebook, pen, methodological instructions, album.

State of health: applicants suffering from acute infectious diseases, including respiratory diseases, are not allowed to attend classes.

A student who is late for class can attend it, but if the teacher has put "ab" in the journal, he must complete it in the general order.

*Use of mobile devices:*

Mobile devices may be used by students with the permission of the teacher if they are needed for the assignment.

*Behavior in the audience:*

The behavior of applicants and teachers in the classrooms must be working and calm, strictly comply with the rules established by the Regulations on academic integrity and ethics of academic relations at Odessa National Medical University, in accordance with the Code of Academic Ethics and University Community Relations of Odessa National Medical University, Regulations on Prevention and detection of academic plagiarism in research and educational work of students of higher education, scientists and teachers of Odessa National Medical University