

SYLLABUS

1. General information on the course

Full course name	Medical Genetics
Full official name of a higher education institution	Sumy State University
Full name of a structural unit	Medical Institute. Department of Pediatrics
Author(s)	Shkolna Iryna Ivanivna, Petrashenko Viktoriia Oleksandrivna
Cycle/higher education level	The Second Level Of Higher Education, National Qualifications Framework Of Ukraine – The 7th Level, QF-LLL – The 7th Level, FQ-EHEA – The Second Cycle
Semester	1 week during 7-th semester
Workload	Discipline in 1 credit ECTS, 30 hours (14 hours is contact work with the teacher, 6 hours of lectures, 10 hours of selfeducation).
Language(s)	English

2. Place in the study programme

Relation to curriculum	Compulsory course available for study programme "Medicine"
Prerequisites	Krok-1, Necessary knowledge of: - Latin language and medical terminology, - medical biology, - human anatomy, - physiology, - histology, cytology and embryology, - biology and immunology, - pathomorphology, - pathophysiology, - pharmacology, - hygiene and ecology, - propedeutics of pediatrics, - radiology.
Additional requirements	There are no specific requirements
Restrictions	There are no specific restrictions

3. Aims of the course

The purpose of the discipline is to achieve students' modern knowledge and professional skills in medica genetic based on a clinical picture of the most common forms of genetic and chromosomal diseases, modern methods of diagnosis and treatment in accordance with the principles of medical ethics and deontology.

4. Contents

<p>Topic 1 Heredity and pathology. The role of heredity in human pathology. Propaedeutics of hereditary pathology</p> <p>Tasks of medical genetics. The role of heredity in human pathology. Syndromological analysis.. Birth defects.</p>
<p>Topic 2 Monogenic diseases</p> <p>General characteristics of monogenic disorders. Clinical pictures of most common monogenic disorders.</p>
<p>Topic 3 Hereditary diseases of metabolism.</p> <p>Treatment of hereditary metabolic diseases based on the principles of evidence-based medicine. Rehabilitation and social adaptation.</p>
<p>Topic 4 Chromosomal disorders</p> <p>General characteristic of chromosomal anomalies (numerical and structural). Clinical pictures of most common chromosomal disorders.</p>
<p>Topic 5 Multifactorial disorders</p> <p>General characteristics of multifactorial disorders. Determination of genetic predisposition. Prevention methods.</p>
<p>Topic 6 Medical-genetic consulting. Methods of prenatal diagnostics. Screening programs. Prevention of hereditary diseases. Medical and genetic counseling. Prenatal diagnosis. Screening programs.</p>
<p>Topic 7 Differential test - medical genetics</p> <p>Differential test - medical genetics</p>

5. Intended learning outcomes of the course

After successful study of the course, the student will be able to:

LO1	Identify risk groups for the development of genetic pathologies. Collect data on patient complaints, anamnesis of a patient with genetic pathology. Interpret the genealogical anamnesis, compile a pedigree. Analyze the type of inheritance or sign of the disease in the family. Carry out selection from the contingent of patients for cytogenetic, biochemical and molecular genetic research methods.
LO2	Evaluate information about the diagnosis based on the results of genetic research methods. Check the most probable diagnosis in the presence of genetic pathology. Prescribe appropriate laboratory and / or instrumental examination in genetic pathology. Carry out differential diagnosis of genetic diseases. Prescribe the necessary regime and treatment for genetic pathology.
LO3	Choose preventive measures to prevent hereditary or congenital diseases. Plan preventive measures to reduce the incidence of the most common multifactorial diseases based on genetic approaches.

6. Role of the course in the achievement of programme learning outcomes

Programme learning outcomes achieved by the course.

For 222 Medicine:

PO1	To detect and identify the leading clinical symptoms and syndromes (according to the List 1); to establish the most probable nosological or syndromic preliminary clinical diagnosis of diseases (according to the List 2) using standard methods, preliminary data of the patient's anamnesis, patient's examination data, and knowledge about a human, his organs and systems.
PO2	To collect information about the patient's general condition; to assess the patient's psychomotor and physical development and the state of organs and systems of the body; to assess information on the diagnosis (according to the List 4) based on laboratory and instrumental findings.
PO3	To order and analyze additional (mandatory and optional) examinations (laboratory, radiological, functional and/or instrumental) (according to the List 4) in order to perform a differential diagnosis of diseases (according to the List 2).
PO4	To establish a final clinical diagnosis at a medical institution under control of a supervising doctor by means of informed decision and logical analysis of the obtained subjective and objective data of clinical and additional examinations, and differential diagnosis, following the relevant ethical and legal norms (according to the List 2).
PO5	To detect the key clinical syndrome or the reason for patient's condition severity (according to the List 3) via informed decision and evaluation of the person's state under any circumstances (at home, in the street, at a healthcare facility), including under emergency and military operation conditions, in the field, with a lack of information and limited time.
PO6	To determine the nature and treatment principles (conservative, operative) in patients with diseases (according to the List 2) at a healthcare facility, at patient's home or during medical evacuation process (including in the field), based on the provisional clinical diagnosis and observing the relevant ethical and legal norms, by making a reasonable decision according to existing algorithms and standard procedures based on the principles of evidence-based medicine; if needed to go beyond the standard scheme, to substantiate the personalized recommendations under control of a supervising doctor at a medical facility.
PO7	To determine an appropriate work and rest mode in the treatment of diseases (according to the List 2) at a healthcare institution, at patient's home and during medical evacuation (including in the field), based on the provisional clinical diagnosis and observing the relevant ethical and legal norms, by making a reasonable decision according to existing algorithms and standard procedures.
PO8	To determine an appropriate diet in the treatment of diseases (according to the List 2) at a healthcare institution, at patient's home and during medical evacuation (including in the field), based on the provisional clinical diagnosis and observing the relevant ethical and legal norms, by making a reasonable decision according to existing algorithms and standard procedures.
PO10	To assess the general condition of a newborn child by making an informed decision according to existing algorithms and standard schemes and adhering to the relevant ethical and legal norms.
PO16	To plan and implement a system of sanitary and preventive measures against the occurrence and spread of diseases among the population.

PO18	To search for the necessary information in the professional literature and databases; to analyze, evaluate, and apply this information. To apply modern digital technologies, specialized software, statistical methods of data analysis to solve complex health problems.
PO19	To assess environmental impact on public health.

7. Teaching and learning activities

7.1 Types of training

<p>Topic 1. Heredity and pathology. The role of heredity in human pathology. Propaedeutics of hereditary pathology</p>
<p>pr.tr.1 "Heredity and pathology. The role of heredity in human pathology. Propaedeutics of hereditary pathology" (full-time course)</p> <p>Tasks of medical genetics. The role of heredity in human pathology. Semiotics of hereditary diseases. Birth defects. The study of this topic involves theoretical work in the classroom with the use of virtual simulation (watching movies) with discussion. Working with genealogical cards.</p>
<p>Topic 2. Monogenic diseases</p>
<p>lect.1 "Monogenic diseases" (full-time course)</p> <p>General characteristics of monogenic pathology. Clinical and genetic characteristics of the most common forms of monogenic diseases. Classes are held in the form of multimedia lectures (for lockdown - online).</p>
<p>pr.tr.2 "Monogenic diseases" (full-time course)</p> <p>General characteristics of monogenic pathology. Clinic and genetics of the most common forms of monogenic diseases. The study of the topic involves theoretical work in the classroom, solving situational tasks, the use of virtual simulation (watching movies about the most common forms of monogenic pathology in population) with further discussion. In the absence of a lockdown, work in hospital departments (according to the agreement on cooperation between hospital and university).</p>
<p>Topic 3. Hereditary diseases of metabolism.</p>
<p>pr.tr.3 "Metabolic disorders" (full-time course)</p> <p>Metabolic disorders. Hereditary metabolic diseases. Treatment of hereditary diseases, which are based on evidence-based medicine. Rehabilitation and social adaptation. The study of topic involves theoretical work in the classroom, solving situational tasks, the use of virtual simulation (watching movies about the most common forms of metabolic diseases) with further discussion. In the absence of a lockdown, work in hospital departments (according to the agreement on cooperation between hospital and university).</p>
<p>Topic 4. Chromosomal disorders</p>

lect.2 "Chromosomal disorders" (full-time course) General characteristics of chromosomal anomalies (numerical and structural). Clinical pictures of most common chromosomal disorders. Classes are held in the form of multimedia lectures (for lockdown - online).
pr.tr.4 "Chromosomal disorders" (full-time course) General characteristics of chromosomal diseases. Clinic and diagnosis of the main forms of chromosomal diseases. The study of topic involves theoretical work in the classroom. In the absence of a lockdown, work in hospital departments (according to the agreement on cooperation between hospital and university). Analysis of karyograms.
Topic 5. Multifactorial disorders
pr.tr.5 "General characteristic of multifactorial disorders. Preventive methods." (full-time course) General characteristics of multifactorial diseases. Determination of genetic predisposition. Prevention measures. The study of topic involves theoretical work in the classroom. In addition, the study of this topic provides role games. In the absence of a lockdown, work with patients in hospital departments (according to the agreement on cooperation between hospital and university).
Topic 6. Medical-genetic consulting. Methods of prenatal diagnostics. Screening programs.
lect.3 "Prenatal diagnostics" (full-time course) Prenatal diagnostics. Screening programs. Classes are held in the form of multimedia lectures (for lockdown - online).
pr.tr.6 "Prenatal diagnostics. Screening programs." (full-time course) Prevention of hereditary diseases. Medical-genetic consulting. Methods of prenatal diagnostics. Screening programs. The study of this topic involves theoretical work in the classroom, using virtual simulation (watching educational movies demonstrating methods of invasive and non-invasive prenatal research) with further discussion. In addition, the study of this topic provides role games to improve skills in diagnosis of birth defects at the prenatal stage.
Topic 7. Differential test - medical genetics
pr.tr.7 "Differential test" (full-time course) Passing complex written differential test

7.2 Learning activities

LA1	Self-study
LA2	Preparation for practical classes
LA3	Interpretation of karyogram, genealogical cards
LA4	Analysis of clinical cases
LA5	Working with a patient in a hospital
LA6	E-learning in different systems (zoom, mix.sumdu.edu.ua)

LA7	Watching educational movies
LA8	Individual research project (article, report on a scientific conference, etc.)
LA9	Work with textbooks and information sources

8. Teaching methods

Course involves learning through:

TM1	Interactive lectures
TM2	Case-based learning (CBL).
TM3	Team-based learning (TBL).
TM4	Research-based learning (RBL).
TM5	Role game
TM6	Brain storm
TM7	Educational discussion / debate

The discipline is taught using modern teaching methods (CBL, TBL, RBL), which promote the development of professional abilities and stimulate creative and scientific activities. The methods are aimed at training practice-oriented specialists.

The discipline provides students with soft skills: GC 1. Ability to abstract thinking, analysis, and synthesis. GC 2. Ability to learn, master modern knowledge, and apply the knowledge in practice. GC 3. Knowledge and understanding of the subject area and professional activity comprehension. GC 4. Ability to adapt and act in a new situation. GC 5. Ability to make reasoned decisions; teamwork ability; interpersonal skills. GC 7. Ability to use information and communication technologies. GC 8. Determination and persistence on the tasks and commitments undertaken.

9. Methods and criteria for assessment

9.1. Assessment criteria

ECTS	Definition	National scale	Rating scale
	Outstanding performance without errors	5 (Excellent)	$170 \leq RD \leq 200$
	Above the average standard but with minor errors	4 (Good)	$140 \leq RD < 169$
	Fair but with significant shortcomings	3 (Satisfactory)	$120 \leq RD < 139$
	Fail – some more work required before the credit can be awarded	2 (Fail)	$0 \leq RD < 119$

9.2 Formative assessment

FA1	Surveys and oral comments on student results
FA2	Solving clinical cases
FA3	Testing

FA4	Peer assessment
FA5	Presentation of an individual research project (presentation at a conference, competition of scientific works)
FA6	Teacher's instructions for practical tasks
FA7	Checking and evaluating written tasks

9.3 Summative assessment

SA1	Evaluation of written works, surveys, solving a clinical case
SA2	Passing complex written differential test
SA3	Presentation of an individual research project (presentation at a conference, competition of scientific works)

Form of assessment:

7 semester	200 scores
SA1. Evaluation of written works, surveys, solving a clinical case	120
	120
SA2. Passing complex written differential test	80
	80

Form of assessment (special cases):

7 semester	200 scores
SA1. Evaluation of written works, surveys, solving a clinical case	120
	In the lockdown, practical classes are held online using the platform mix.sumdu.edu.ua, Zoom, Google Meet.
	120
SA2. Passing complex written differential test	80
	In the lockdown, practical classes are held online using the platform mix.sumdu.edu.ua, Zoom, Google Meet.
	80

At each practical class, student receives a point on a four-point scale, where "5" - excellent, "4" - good, "3" - satisfactory, "2" - unsatisfactory. At the end of the discipline, the points are summed and the average is calculated, which is converted on a 120-point scale. A student may take a complex differential test if he has fulfilled the requirements of the curriculum and received at least 72 points for the current educational activity. Differential credit is made according to the schedule at the end of the discipline. The grade for modular control is set in the traditional 4-point grading system with subsequent conversion on an 80-point scale, with a grade of "5" corresponds to 80 points, "4" - 64 points, "3" - 48 points, "2" - 0 points. Points for current performance and comprehensive differential test are summed. The total score > 120 is considered to be the successful completion of the discipline. Differential test is carried out according to the schedule at the end of the discipline. Points for differential complex test set in a 4-point system and is converted on an 80-point scale (this grade "5" corresponds to 80 points, "4" - 64 points, "3" - 48 points, "2" - 0 points). Then points are summed up. The total score > 120 is considered to be the successful completion of the discipline.

10. Learning resources

10.1 Material and technical support

MTS1	Information and communication systems
MTS2	Library materials, archive of karyograms, genealogical cards, results of biochemical screening blood tests.
MTS3	Computers, computer systems and networks
MTS4	Non-profit municipal enterprise of Sumy regional council "Regional Children's Clinical Hospital"
MTS5	Multimedia, video equipment, projection equipment (video cameras, projectors, laptop screens)

10.2 Information and methodical support

Essential Reading	
1	Medical Genetics : study guide / V. O. Petrashenko, A. M. Loboda, S. M. Kasian ; under the editorship of S. V. Popov. – Sumy : Sumy State University, 2018. –140 p.
2	USMLE Step 1: Biochemistry and Medical Genetics [Текст] : Lecture Notes / Editors S. Turco, R. Lane, R.M. Harden. — New York : Kaplan, 2019. — 409 p.
3	Nelson Textbook of Pediatrics [Text] / R. M. Kliegman [et al.] ; ed. R. E. Behrman. - 21th ed. - Edinburgh [etc.] : Elsevier, 2020.
Supplemental Reading	
1	Modern methods of genetic diagnosis [Текст] : study guide / V. E. Markevich, V. O. Petrashenko, O. K. Redko etc. — Sumy : Sumy State University, 2015. — 214 p.
2	Neonatology. Introduction [Текст]: study guide/ O.K. Redko, V.O. Petrashenko, I.V. Tarasova, I.E. Zaitsev. - Sumy: Sumy State University, 2017. - 182 p. - ISBN 978-966-657-677-7
3	Pediatrics: textbook / O. V. Tiazhka, T. V. Pochinok, A. M. Antoshkina etc. ; edited by O.V. Tiazhka. — 3-rd edition, reprint. — Vinnytsia : Nova Knyha, 2018. — 544 p
4	The Standards of Practical Skills in Neonatology [Текст]: study guide / Ye.Ye. Shunko, A.M. Loboda, I.V. Tarasova [et al.]. - Sumy: Sumy State University, 2018. - 315 p.
Web-based and electronic resources	
1	https://ocw.sumdu.edu.ua/content/1006