# COURSE PROGRAM

## SYLLABUS

### 1. General information on the course

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| **Full course name** | Medical Genetics |
| **Full official name of a higher education institution** | Sumy State University |
| **Full name of a structural unit** | Academic and Research Medical Institute. Кафедра педіатрії |
| **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 |
| **Duration** | one semester |
| **Workload** | 1 ECTS, 30 hours. For full-time course 22 hours are working hours with the lecturer (4 hours of lectures, 18 hours of seminars), 8 hours of the individual study. |
| **Language(s)** | English |

### 2. Place in the study programme

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| **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

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| Topic 1 Heredity and pathology. The role of heredity in human pathology. Propaedeutics of hereditary pathology<br>Tasks of medical genetics. The role of heredity in human pathology. Syndromological analysis.. Birth defects. |
| Topic 2 Monogenic diseases<br>General characteristics of monogenic disorders. Clinical pictures of most common monogenic disorders. |
| Topic 3 Metabolic diseases<br>General characteristics of hereditary metabolic diseases. Clinical characteristics of some forms of hereditary metabolic diseases. |
| Topic 4 Hereditary diseases of metabolism<br>Treatment of hereditary metabolic diseases based on the principles of evidence-based medicine. Rehabilitation and social adaptation. |
| Topic 5 Chromosomal disorders<br>General characteristics of chromosomal diseases. Etiology, cytogenetics, clinical characteristics of some forms of chromosomal diseases |
| Topic 6 Chromosomal disorders<br>Principles of rehabilitation and social adaptation of patients with chromosomal pathology. |
| Topic 7 Multifactorial disorders<br>General characteristics of multifactorial disorders. Determination of genetic predisposition. Prevention methods. |
| Topic 8 Medical-genetic consulting. Methods of prenatal diagnostics. Screening programs<br>Prevention of hereditary diseases. Medical and genetic counseling. Prenatal diagnosis. Screening programs. |
| Topic 9 Differential test - medical genetics<br>Differential test - medical genetics |

## 5. Intended learning outcomes of the course

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

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| LO1 | Identify risk groups for the development of genetic pathologies. Collect data on patient complaints, anamnesis morbi, anamnesis vitae of a patient with genetic pathology. |
| LO2 | Assess diagnostic information using a standard procedure based on the results of genetic testing. Prescribe an appropriate laboratory and/or instrumental examination in the presence of genetic pathology. Carry out differential diagnosis of genetic diseases. |

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| LO3 | Conduct selection from the contingent of individuals for cytogenetic, biochemical and molecular genetic research methods. |
| LO4 | Determine the necessary mode of study, work and rest of children in the treatment of genetic pathology. Plan preventive measures aimed at preventing the occurrence of hereditary or congenital diseases. To plan preventive measures to reduce the frequency of the most common diseases of multifactorial genesis based on genetic approaches. |
| LO5 | Determine the nutrition of children during the treatment of hereditary diseases. |
| LO6 | Determine the necessary regimen and volume of treatment in the presence of genetic pathology. |
| LO7 | To acquire counseling skills on family planning and medical and genetic counseling. |
| LO8 | To determine the management tactics of children with genetic pathology, who are subject to dispensation |
| LO9 | Assess the impact of the environment on children's health and the risk of developing hereditary and/or congenital pathology |
| LO10 | Work with professional literature, analyze and use the information received. Use professional vocabulary in practical activities. |
| LO11 | Collect genealogical anamnesis, compile a pedigree, analyzing the type of inheritance or a sign of illness in the family. |
| LO12 | To acquire the skills of clinical examination of children of different age groups with a genetic predisposition to the development of hereditary diseases. |

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

Programme learning outcomes achieved by the course.
For 222 Medicine:

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| 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). |

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| 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. |
| PO14 | To perform medical procedures (according to the List 5) at a medical facility, at home or at work on the basis of a provisional clinical diagnosis and/or health parameters through making an informed decision and adhering to the relevant ethical and legal norms. |
| 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. Soft Skills

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| SS1 | Ability to abstract thinking, analysis and synthesis. |
| SS2 | Ability to learn and acquire modern knowledge |
| SS3 | Knowledge and understanding of the subject area and understanding of professional activity |
| SS4 | Ability to adapt and act in a new situation |
| SS5 | Ability to make informed decisions; work in a team; interpersonal skills. |
| SS6 | Ability to use information and communication technologies |

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| SS7 | Determination and persistence in relation to assigned tasks and assumed responsibilities. |

## 8. Teaching and learning activities

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| **Topic 1. Heredity and pathology. The role of heredity in human pathology. Propaedeutics of hereditary pathology** |
| pr.tr.1 "Heredity and pathology. The role of heredity in human pathology." (full-time course)<br>asks 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. |
| **Topic 2. Monogenic diseases** |
| lect.1 "Monogenic diseases" (full-time course)<br>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). |
| pr.tr.2 "Monogenic diseases" (full-time course)<br>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). |
| **Topic 3. Metabolic diseases** |
| pr.tr.3 "Metabolic diseases" (full-time course)<br>Hereditary metabolic diseases. Principles of treatment of hereditary diseases based on evidence-based medicine. Rehabilitation and social adaptation. The study of this topic involves theoretical work in the classroom, solving situational problems, using virtual simulation (watching films about the most common variants of metabolic diseases) with further discussion. If possible, work at the patient's bedside in the departments of the medical institution (according to the cooperation agreement between the medical institution and the university). |
| **Topic 4. Hereditary diseases of metabolism** |
| pr.tr.4 "Metabolic disoders" (full-time course)<br>Metabolic disoders. Principles of treatment of hereditary diseases based on evidence-based medicine. Rehabilitation and social adaptation. The study of this topic involves theoretical work in the classroom, solving situational problems. If possible, work at the patient's bedside in the departments of the medical institution (according to the cooperation agreement between the medical institution and the university). |
| **Topic 5. Chromosomal disorders** |

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| lect.2 "Chromosomal disorders" (full-time course)<br>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.5 "Chromosomal diseases" (full-time course)<br>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 6. Chromosomal disorders** |
| pr.tr.6 "Chromosomal disorders" (full-time course)<br>Principles of rehabilitation and social adaptation of patients with chromosomal pathology. The study of this topic involves theoretical work in the classroom. In the absence of quarantine restrictions, work at the patient's bedside in specialized departments of the medical institution (according to the agreement on cooperation between the medical institution and the university) |
| **Topic 7. Multifactorial disorders** |
| pr.tr.7 "General characteristics of multifactorial disorders. Preventive methods" (full-time course)<br>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 8. Medical-genetic consulting. Methods of prenatal diagnostics. Screening programs** |
| pr.tr.8 "Prenatal diagnostics" (full-time course)<br>Levels and ways of prevention of hereditary diseases. Medical and genetic counseling. Prenatal diagnosis. Screening programs. The study of this topic involves theoretical work in the classroom, the use of virtual simulation (watching films demonstrating the methods of conducting invasive and non-invasive prenatal research) with further discussion. Interpretation of double and triple biochemical screening results and fetal ultrasound analysis. In addition, when studying this topic, role-playing games are provided to improve the skills of timely diagnosis of developmental defects at the prenatal stage. |
| **Topic 9. Differential test - medical genetics** |
| pr.tr.9 "Differential test - medical genetics" (full-time course)<br>Passing complex written differential test |

## 9. Teaching methods

9.1 Teaching methods

Course involves learning through:

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| TM1 | Case-based learning |

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| TM2 | Team Based Learning |
| TM3 | Research Based Learning |
| TM4 | Practical training |
| TM5 | Self-study |
| TM6 | Electronic learning |
| TM7 | Lecture teaching |

The discipline is taught using modern teaching methods (CBL, TBL, RBL), which contribute not only to the development of professional abilities, but also encourage creative thinking

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9.2 Learning activities

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| LA1 | Preparation for practical classes |
| LA2 | Analysis of clinical cases |
| LA3 | Practical work with the patient in specialized departments of the hospital |
| LA4 | Electronic learning in systems (Zoom, MIX.sumdu.edu.ua) |
| LA5 | Individual research project (student research paper, article, theses, etc.) |
| LA6 | Working with textbooks and relevant information sources |
| LA7 | Performance of group practical task |
| LA8 | Interpretation of karyograms |
| LA9 | Interactive lectures |

## 10. Methods and criteria for assessment

10.1. Assessment criteria

| Definition | National scale | Rating scale |
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| 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) | $0 \leq RD < 119$ |

10.2 Formative assessment

| | **Description** | **Deadline, weeks** | **Feedback** |
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| FA1<br>Current evaluation of the level of theoretical and practical training | Includes oral interview, interpretation of kariograms, laboratory and instrumental methods of examination, objective structured examination of the patient, solution of clinical group and individual cases, ongoing testing. Students who are involved in research activities have the opportunity to present the results of their own research at conferences, student research competitions, etc. (incentive activities, additional points) | During the entire period of studying the discipline | Held at each class, the result of performing the LA affects the comprehensive assessment for the practical class |
| FA2<br>Teaching advice during the preparation of an individual research project (speech at a conference, competition of scientific works) | An important factor in the formation of professional qualities of future specialists is the research work of students. Involvement of students in research activities contributes to the formation of their scientific worldview, industriousness, work capacity, initiative, etc. | During the entire period of studying the discipline | Teacher's oral comments. The student is given additional incentive points (from 5 to 10), depending on the type of research project |
| FA3<br>Instructions of the teacher in the process of performing practical tasks | The guidelines reveal the methods of pedagogical control over the professional activities of applicants. Efficiency is determined by compliance with all stages of practical tasks. | During the entire period of studying the discipline | Counseling of students in working with a standardized patient, direct and indirect observation of the work of the examiners "at the bedside" of the patient with subsequent determination of the level of practical training |

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| FA4<br>The survey and the teacher's oral comments on his results | This provides an opportunity to identify the state of educational experience acquired by students in accordance with the set goals, to find out the prerequisites for the state of formation of the obtained results, the causes of difficulties, to adjust the learning process, to track the dynamics of the formation of learning results and prognosis their development. | During the entire period of studying the discipline | According to the obtained data on the results of training, based on their analysis, it is proposed to determine the evaluation as an indicator of the achievements of the educational activities of the applicants |
| FA5<br>Processing of clinical cases | The case method makes it possible to reveal and form the qualities and abilities of medical students necessary for further work, forms clinical thinking, analytical abilities, independence in decision-making, communication, skills for working with a sufficiently large amount of information. | During the entire period of studying the discipline | Assessment of the student's ability to think clinically, justify their decisions, clearly express their opinions, determine the level of theoretical training, which is reflected in the corresponding assessment |
| FA6<br>Tests (automated tests) to control the educational achievements of applicants | A method of effective verification of the level of assimilation of knowledge, abilities and skills from each subject of an educational discipline. Testing allows you to check the assimilation of educational material from each subject. | During the entire period of studying the discipline | The student must provide 60% of the correct answers, which is an admission to the practical part of the lesson |

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| FA7<br>Task of assessing the level of theoretical training | Assessment of theoretical knowledge from the subject of the discipline. It is conducted at each practical session in accordance with the specific goals of each topic on the basis of a comprehensive assessment of activity | During the entire period of studying the discipline | Feedback is aimed at supporting students' independent work, identifying shortcomings and assessing the level of acquired theoretical knowledge |
| FA8<br>Focus group discussions | The method makes it possible to involve all participants in the process of discussion and justification of one's own opinion through multilateral communication, to develop the ability to conduct a professional discussion, to cultivate respect for colleagues and the ability to generate alternative ideas and proposals. | During the entire period of studying the discipline | Assessment of the student's ability to work in a team, ability to justify their decisions, determination of the level of theoretical training, which is reflected in the corresponding assessment |
| FA9<br>Peer assessment | Partnership interaction aimed at improving the results of educational activities by comparing one's own current level of success with previous indicators. Provides an opportunity to analyze one's own educational activities | During the entire period of studying the discipline | Adjustment of approaches to learning together with students, taking into account the results of the assessment |
| FA10<br>Tests (automated tests) to control the educational achievements of applicants | A method of effective verification of the level of assimilation of knowledge, abilities and skills from each subject of an educational discipline. Testing allows you to check the assimilation of educational material from each subject. | During the entire period of studying the discipline | The student must provide 60% of the correct answers, which is an admission to the practical part of the lesson |

10.3 Summative assessment

| | Description | Deadline, weeks | Feedback |
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| SA1<br>Current evaluation of the level of theoretical and practical training | Includes oral interview, interpretation of laboratory and instrumental methods of examination, objective structured examination of the patient, solution of clinical group and individual cases, ongoing testing. Students who are involved in research activities have the opportunity to present the results of their own research at conferences, student research competitions, etc. (incentive activities, additional points) | During the entire period of studying the discipline | Carrying out the results of the LA in each lesson affects the comprehensive assessment for the practical lesson |
| SA2<br>Differentiated assessment | Students who have successfully mastered the material can be admitted to the module | In the last class in discipline | A student can get 80 points for a differential credit. The minimum number of points a student must r |
| SA3<br>Final testing | A method of effective verification of the level of assimilation of knowledge, abilities and skills in an educational discipline. Testing allows you to check the results of training during the cycle and determine the level of knowledge at the end of the discipline. | Final computer test at the end of the course (10 points) | It is an admission to the module |

Form of assessment:

| | | Points | Можливість перескладання з метою підвищення оцінки |
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| **The first semester of teaching** | | **200 scores** | |
| SA1. Current evaluation of the level of theoretical and practical training | | **110** | |
| | | 110 | No |
| SA2. Differentiated assessment | | **80** | |
| | | 80 | No |
| SA3. Final testing | | **10** | |
| | | 10 | No |

Points from the discipline, defined as the sum of points for the current educational activity (at least 72) and points for the final module control (at least 48). Points for the current activity are calculated according to the formula 110xthe arithmetic average of the student's performance in the 4-point evaluation system / 5. The student receives a maximum of 10 points for diagnostic testing. The minimum number of points that a student must receive is 6 points. The maximum number of points for the current educational activity of the student is 120. The student is admitted to the credit provided that the requirements of the educational program are met and if he has scored at least 72 points for the current educational activity: 66 points during practical classes and 6 points for testing.

The final module control is conducted at the end of the academic semester in the form of a written assessment, while the grade "5" corresponds to 80 points, "4" - 64 points, "3" - 48 points, "2" - 0 points. In case of an unsatisfactory result for the final module control, the student has the right to retake the assessment. Students who did not appear for the assessment without a good reason are considered to have received an unsatisfactory grade. A student's refusal to complete the final module task is certified as an unsatisfactory response. Incentive points are added to the grade in the discipline for the implementation of an individual research project (defense of a student thesis 10 points, speech at a conference, poster presentation at a conference, theses of reports - 5 points). The total score for the discipline cannot exceed 200 points. The possibility of re-crediting the points obtained under the system of non-formal education is provided in accordance with the Regulations.

## 11. Learning resources

11.1 Material and technical support

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| MTS1 | Information and communication systems, software (to support distance learning) |
| MTS2 | Library funds, archive of kariograms, genealogical cards, results of biochemical screening blood tests (double test, triple test) and ultrasound screening |
| MTS3 | Computers, computer systems and networks |
| MTS4 | Municipal non-profit enterprise of Sumy regional council "Regional children's clinical hospital" |
| MTS5 | Multimedia, video and sound reproduction, projection equipment (video cameras, projectors, laptop screens) |

11.2 Information and methodical support

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| **Essential Reading** | |
| 1 | USMLE Step 1: Biochemistry and Medical Genetics [Текст] : Lecture Notes / Editors S. Turco, R. Lane, R.M. Harden. — New York : Kaplan, 2019. — 409 p. |
| **Supplemental Reading** | |
| 1 | Medical Genetics [Текст] : 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 | 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 |
| 3 | 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 |
| 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/796 |

# COURSE DESCRIPTOR

| № | Course Bescriptor | Total hours | Classroom work, hours | | | | Independent work of students, hours | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | Total hours | Lectures | Workshops (seminars) | Labs | Total hours | Self-study of the material | Preparation for workshops (seminars) | Preparation for labs | Preparation for assesment | Independent extracurricular tasks | |
| 1 | 2 | | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 | 11 | 12 | 13 |
| **full-time course** | | | | | | | | | | | | | |
| 1 | Heredity and pathology. The role of heredity in human pathology. Propaedeutics of hereditary pathology | | 2.5 | 2 | 0 | 2 | 0 | 0.5 | 0 | 0.5 | 0 | 0 | 0 |
| 2 | Monogenic diseases | | 5 | 4 | 2 | 2 | 0 | 1 | 0.5 | 0.5 | 0 | 0 | 0 |
| 3 | Metabolic diseases | | 2.5 | 2 | 0 | 2 | 0 | 0.5 | 0 | 0.5 | 0 | 0 | 0 |
| 4 | Hereditary diseases of metabolism | | 2.5 | 2 | 0 | 2 | 0 | 0.5 | 0 | 0.5 | 0 | 0 | 0 |
| 5 | Chromosomal disorders | | 5 | 4 | 2 | 2 | 0 | 1 | 0.5 | 0.5 | 0 | 0 | 0 |
| 6 | Chromosomal disorders | | 2.5 | 2 | 0 | 2 | 0 | 0.5 | 0 | 0.5 | 0 | 0 | 0 |
| 7 | Multifactorial disorders | | 2.5 | 2 | 0 | 2 | 0 | 0.5 | 0 | 0.5 | 0 | 0 | 0 |
| 8 | Medical-genetic consulting. Methods of prenatal diagnostics. Screening programs | | 2.5 | 2 | 0 | 2 | 0 | 0.5 | 0 | 0.5 | 0 | 0 | 0 |
| 9 | Differential test - medical genetics | | 2.5 | 2 | 0 | 2 | 0 | 0.5 | 0 | 0.5 | 0 | 0 | 0 |
| Assesment | | | | | | | | | | | | | |
| 1 | Graded Credit | | 6 | 0 | 0 | 0 | 0 | 6 | 0 | 0 | 0 | 6 | 0 |
| Independent extracurricular tasks | | | | | | | | | | | | | |
| *Total (full-time course )* | | | *30* | *22* | *4* | *18* | *0* | *8* | *1* | *4.5* | *0* | *6* | *0* |