

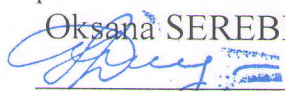
National Pirogov Memorial Medical University, Vinnytsya

"APPROVE"

Vice -Rector of higher education institution

for Research, Education and Teaching professor of HEI

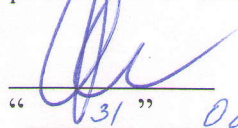
Oksana SEREBRENNIKOVA



" 31 " 08 2022 year

" AGREED "

Head of department of Pediatric №2 professor of HEI Veronika DUDNYK



" 31 " 08 2022 year

SYLLABUS

**OF ACADEMIC DISCIPLINE
(elective course)
«PEDIATRIC ORPHAN DISEASES»**

| | |
|-----------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Specialty | 222 Medicine |
| Educational level | the second (master's) level |
| Educational programme | EPP Medicine, 2022 |
| Academic year | 2022-2023 |
| Department | <i>Pediatric №2</i> |
| Contact information | <i>pediatric2@vnmue.edu.ua, 108, Khmelnytske shosse, Vinnytsya, 21029 Communal Nonprofit Enterprise "Vinnytsia Regional Children's Clinical Hospital of Vinnytsia Regional Council", Tel (0432)560819</i> |
| Syllabus compiler | Associate professor of HEI, PhD Maryna SHALAMAI |

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1. Status and structure of the discipline

| | |
|-------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------|
| Discipline status | elective |
| Discipline code in EPP/discipline place in EPP | EC 5.15, discipline of professional training |
| Course / semester | 5 th year (IX-X semesters) |
| The amount of discipline (the total number of hours / number of credits ECTS) | 60 hours /2 credit ECTS |
| Number of content modules | 1 module |
| The structure of the discipline | Lectures 20 hours Practical classes 10 hours Independent work 30 hours In total: practical classes – 50%, individual student`s work – 50% |
| Language of study | English |
| Form of study | Full-time (<i>or remote full-time by order</i>) |

2. Description of the discipline

Short annotation of the course, relevance. The subject area of the discipline is to obtain knowledge of the mandatory discipline of professional training, the study of which is necessary for the successful professional activity. According to modern knowledge, any human pathology is more or less related to heredity. This provision is the basis for teaching and studying pediatric orphan disorders as a clinical and preventive discipline. Because heredity and variability are integral parts of life, genetics should be the basis of theoretical and clinical training of physicians. 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 understand the use of theoretical knowledge of genetics in clinical practice, replenishment of knowledge in medical and clinical genetics, especially modern problems of diagnosis, treatment and prevention of hereditary pathology, the study of a number of "new" common clinical forms of hereditary diseases.

Prerequisites. For successful mastering of discipline «Pediatric orphan diseases» is based on general natural disciplines: medical biology and genetics, medical and biological physics, medical chemistry, bioorganic chemistry, morphology disciplines, normal and pathological physiology, propedeutics of internal and pediatric diseases and integrates in this disciplines. Lays the foundations for students to study modern diagnostic technologies used in the diagnosis of orphan diseases and used in general clinical practice, lays an understanding of modern

features of rare childhood diseases, as well as common human diseases that occur on the background of hereditary predisposition and require integration of classical clinical ideas and modern high technology, which involves the integration of teaching with different disciplines and the formation of skills to apply knowledge about orphan diseases in the process of further study and professional activities.

The purpose of the course and its significance for professional activities.
The aim of the discipline is to improve theoretical knowledge and practical skills for 5th course students in matters of early diagnosis, principles of treatment of orphan diseases.

Postrequisites. In the process of studying the discipline obtain the knowledge which will be necessary for successful mastering of disciplines, namely: pediatrics, pediatric infectious diseases, internal medicine, and other clinical disciplines, which involve the integration of teaching with these disciplines and the application of knowledge, skills and abilities in the process training and professional activities. Students study the discipline in the form of differential diagnosis of the most common orphan diseases of childhood, practical skills of diagnosis and treatment of emergencies on the basis of stimulation class.

3. Learning outcomes.

After successful study of the discipline the applicant will be able to:

1. Know the basics of information and communication technologies and be able to use them in professional activities.
2. Collect data on patient complaints, medical history, family history, conduct a physical examination of the patient, be able to make and analyze the patient's pedigree.
3. Highlight the leading clinical symptom or syndrome.
4. Establish the most probable diagnosis.
5. Assign laboratory and / or instrumental examinations, analyze the results.
6. Carry out differential diagnosis of hereditary diseases, using the most probable or syndromic diagnosis, data of laboratory and instrumental examination of the patient.
7. Establish a preliminary clinical diagnosis of hereditary disease using knowledge and logical analysis.
8. Determine the necessary therapeutic nutrition in the treatment of orphan diseases.
9. To determine the principles and nature of treatment of patients with hereditary pathology.
10. Perform counseling about family planning.

4. Content and logistic of the discipline

| | | |
|--------------------------------------|---------------------------------------|---------------------------------------------------------------------------------------|
| Module 1 «Pediatric orphan diseases» | 9 or 10 semester 60 hours/2 credit | Practical classes № 1-5 Topics for self- study № 2.1, 3.1, 3.2, 4.1, 4.2, 5.1, 5.2 |
|--------------------------------------|---------------------------------------|---------------------------------------------------------------------------------------|

The course includes 5 topics, which are part of 1 thematic module.

Lectures

1. Rare inborn errors of metabolism 1
2. Rare inborn errors of metabolism 2
3. Rare endocrine disorders in children
4. Rare blood and hematopoietic system disorders. Coagulopathies. Trombocytopatiya.
5. Rare blood and hematopoietic system disorders. Hemoglobinopathies.
6. Rare skin disorders.
7. Rare connective tissue disorders.
8. Rare diseases of the circulatory system
9. Rare inborn malformations
10. Rare disorders of nervous system

The topics of the lecture course reveal the problematic issues of the relevant sections of the discipline.

Module 1. «Pediatric orphan diseases»

Topic 1. General information about orphan diseases. Semiotics of rare diseases.

Topic 2. Examination of a patient with orphan disease.

ISW 2.1 Rare disorders of skin

Topic 3. Rare metabolic disorders. Clinical case of Gaucher disease and mucopolysaccharidosis.

ISW 3.1 Lysosomal storage diseases.

ISW 3.2 Rare diseases of the endocrine system in children.

Topic 4. Rare diseases of the blood. Clinical case of coagulopathies.

ISW 4.1 Trombocytopatiya. Hemoglobinopathies.

ISW 4.2 Rare diseases of the circulatory system

Topic 5. Rare inborn malformations

ISW 5.1 Rare muscular-skeletal, connective tissue disorders.

ISW 5.2 Rare disorders of nervous system

Practical classes provide a theoretical justification of the main issues of the topic and the learning of the following practical skills:

- 1) to examine patients and their relatives and identify congenital and hereditary pathology;

- 2) to determine the clinical features of hereditary pathology and the condition of patients;
- 3) to evaluate the diagnostic, prognostic value of the detected symptoms and morphogenetic variants (microanomalies of development);
- 4) correctly collect genetic history, make a pedigree, determine the type of inheritance;
- 5) to diagnose the most common forms of hereditary pathology.

During practical classes, students in workbooks formulate conclusions on the topic and discuss clinical case, write pedigrees and analyze results of karyotyping.

The student's independent work involves preparation for practical classes and development of practical skills, study of topics for independent extracurricular work, elaboration of scientific literature and writing reviews according to recommended topics for individual work. The control of studying the topics of independent work is carried out on the final module.

Calendar plans of practical classes, thematic plan of independent extracurricular work, volume and directions of individual work are published on the website of the department.

Route of receiving materials: Department of Pediatrics №2 / Student / Full-time / Medicine / 5th year / Educational and methodical materials.

Access to materials is provided from the student's corporate account s000XXX@vnmueu.ua.

5. Forms and methods of monitoring academic

| | |
|-----------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Current control in practicals | Methods: oral or written survey, electronic survey, solving situational problems, working with pedigree diagrams and karyograms, interpretation and evaluation of results (registration of the protocol in the workbook) |
| Final semester control (Credit) at the end of 9 or 10 semesters | According to the Regulation of the Academic process in National Pirogov Memorial Medical University (link https://www.vnmueu.ua/General information) |
| Learning success diagnostic tools | Theoretical questions, tests, clinically-oriented situational tasks, pedigrees, results of karyotyping. |

6. Assessment criteria

Knowledge assessment is carried out in accordance with the Regulations of the Academic process in National Pirogov Memorial Medical University (link <https://www.vnmueu.ua/General> information)

| | |
|-----------------|------------------------------------------------------------------------------------------------------------------|
| Current control | On a four point system of traditional assessments: 5 «excellent», 4 «good», 3 «satisfactory», 2 «unsatisfactory» |
|-----------------|------------------------------------------------------------------------------------------------------------------|

| | |
|-----------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Control of practical skills | According to the four-point system of traditional assessments |
| Credit | 200-points score (average mark converts into points) Credited: 120 to 200 points Not credited: less than 120 points (See Grading Scale) |
| Discipline assessments: | On a 200-points scale (the arithmetic average grade for the semester is converted into points) Credited: 120 to 200 points Not credited: less than 120 points (See Grading Scale) Individual student`s work – 8 to 12 points Total 122 to 200 points |

Discipline Score Scale: National and ECTS

| The sum of grades for all types of educational activities | Score ECTS | Score on a national scale | |
|-----------------------------------------------------------|------------|-------------------------------------------------------------|------------------------------------------------------------|
| | | For exam, course project (work), practice | for credit test |
| 180-200 | A | Excellent | credited |
| 170-179,9 | B | Good | |
| 160-169,9 | C | | |
| 141-159,9 | D | Satisfactory | |
| 122-140,99 | E | Satisfactory | - |
| 0-121,99 | FX | unsatisfactory with the possibility of reassembly | is not credited with the possibility of reassembling |
| | F | unsatisfactory with a mandatory reexamination of discipline | is not credited with mandatory reexamination of discipline |

Criteria for student knowledge assessment

Assessment of oral / written answers during the current assessment

The grade **"excellent"** is given to a student who has deeply and comprehensively mastered the theoretical material, competently and logically explain it. He is fluent in Latin terminology, clearly answers non-standard questions on the topic of the lesson, is able to link the material of the topic with previously studied sections, which indicates knowledge of the recommended literature and the ability to analyze the material studied, and clearly demonstrates the importance of theoretical knowledge for practice medicine.

The grade **"good"** is given to a student who knows and has a good theoretical material, teaches it correctly, does not allow inaccuracies in the answer, is able to reveal the topic from the standpoint of its medical significance and practical application, but the answers do not go beyond the textbook, guidelines.

A grade of **"satisfactory"** is given to a student who knows the basic concepts and definitions of the studied topic, but admits significant inaccuracies or has difficulty in formulating the answer, does not understand the medical aspects of the topic, can not relate theoretical material to practice.

The **grade "unsatisfactory"** is given to a student who does not know the theoretical foundations of the topic, makes gross mistakes in answering, does not understand the basic concepts and definitions, can not explain the importance of theoretical material for practical medicine.

Assessment of practical skills during the current assessment

The grade **"excellent"** is given to a student who knows the course and sequence of independent learning to perform a practical task, seeks the best options for diagnosis, and demonstrates the correct implementation of the necessary practical skills, and true with clear formulations of conclusions.

A grade of **"good"** is given to a student who makes inaccuracies in describing the phenotype, but is able to identify errors and can demonstrate the implementation of practical skills in general, accurately draws up the results of the pedigree in the notebook.

Assessment of **"satisfactory"** is given to a student who knows the basics of the practical task, but has difficulty in performing this, cannot demonstrate the correct sequence of practical skills to describe the phenotype, fully interpret the results of examinations, make pedigree.

The grade **"unsatisfactory"** is given to a student who cannot demonstrate practical skills, has significant difficulties in describing the phenotype, compiling a pedigree and determining the type of inheritance, karyogram analysis, does not register the progress of work in the protocol.

Assessment of independent student work

The student's independent work is assessed during the final classes of the module of the discipline through an oral interview on topics that are not included in the plan. Evaluation of prepared messages, presentations on the selected topic is carried out by traditional evaluation.

The grade **"excellent"** is given to a student who has deeply and comprehensively revealed the problem, logically stated the main issues, give examples from medical informative sources. He is able to connect the material of this topic with the previously studied sections, which indicates the ability to analyze the studied material, as well as clearly demonstrates the importance of the acquired theoretical knowledge for practical medicine.

The grade **"good"** is given to a student, who knows and is well versed in theoretical material, competently revealed the main issues of the topic and its medical significance, but did not go beyond the textbook, guidelines.

A grade of **"satisfactory"** is given to a student who has revealed the basic concepts and definitions of the recommended topic, but has not fully disclosed it,

does not understand the medical aspects of the topic, relate theoretical material to practice.

The calculation of individual points is carried out on the basis of the Regulation of the Academic process in National Pirogov Memorial Medical University (Вінниця, 2020). (link <https://www.vnmu.edu.ua/General> information).

12 points - added to the assessment of the discipline for a student who won a prize at the interuniversity competitions in the discipline or a prize at the Ukrainian competition of student research reports or a prize at the interuniversity / international scientific conference with the published work;

11 points - are added to the assessment of the discipline for a student who won the first prize at the intra-university Olympiad in the discipline

10 points - are added to the assessment of the discipline for a student who won a prize (II-III) at the intra-university Olympiad in the discipline or at the student scientific conference with the availability of printed work; or for participation (without a prize place) in interuniversity competitions in the discipline or a prize place in an interuniversity / international scientific conference with the availability of published work.

9 points - are added to the assessment of the discipline for a student who participated (without a prize) in the intra-university Olympiad in the discipline or student scientific conference with the presence of published work

8 points - are added to the assessment of the discipline for a student who actively participated in the student scientific group, published a paper with results of scientific and practical research, but did not personally participate in the student scientific conference, prepared a poster report.

Evaluation of final control (credit)

The final control is credited if the student has mastered totally the sections of the discipline confirmed by the current assessment of each practical lesson and has attended a lecture course. To assess the final control the calculation of the arithmetic mean of traditional grade for the semester is calculated.

7. Policy of discipline / course

The student has the right to receive high-quality educational services, access to contemporary scientific and educational information, qualified tutoring during the study of discipline and mastering practical skills. The policy of the department during the providing of educational services is a student-centered, based on normative documents of the Ministry of Education and the Ministry of Health of Ukraine, the Statute of the University and the Procedure for the Providing of Educational Services regulated by the main principles of the organization of the educational process in National Pirogov Memorial Medical University and the principles of academic integrity

Adherence to the rules of National Pirogov Memorial Medical University, safety techniques in practical classes.

Occupational safety instruction with students during labor, professional training in health care institutions is conducted by the head of the structural unit of the institution or the student's immediate supervisor. Before the beginning of practical

classes in the first lesson (at the beginning of each semester) the teacher, according to the requirements of the Job description, conducts instruction on labor protection (including rules of personal hygiene, sanitary regime, danger during supervision of patients, etc.). Targeted instruction is conducted with students in the case of organization outside of educational activities (competitions, conferences, etc.).

Requirements for preparation for practical classes. The student must be prepared for a practical lesson, situational tasks for the current topic should be solved in a workbook.

They should come to class on time, without delay. A student who is late more than 10 minutes for class is not allowed to be present and must rework it in the prescribed manner.

In practical classes, the student must be dressed in a work uniform (medical gown, hat, medical mask, changeable shoes). Students who do not have a work uniform are not allowed to study.

The student must follow safety rules during practical classes and in the hospital.

When discussing theoretical issues, students should demonstrate tolerance and respect for their colleagues and the teacher.

Usage of mobile phones and other electronic devices.

The use of mobile phones and other electronic devices in the classroom is allowed only during electronic testing or surveys.

Academic integrity. When studying the discipline, the student must be guided by the Code of Academic Integrity and Corporate Ethics of National Pirogov Memorial Medical University (link: <https://www.vnmdu.edu.ua/General> information)/ Code of Academic Integrity). In case of violation of the norms of academic integrity during the current and final controls student receives a grade of "2" and must work it out to his teacher in the prescribed manner within two weeks after receiving an unsatisfactory assessment).

Missed classes. Missed classes are reworked in the manner prescribed by the Regulations on the organization of the educational process in National Pirogov Memorial Medical University (link <https://www.vnmdu.edu.ua/General> information / Basic documents) at the time specified in the schedule (published on the website of the department <https://www.vnmdu.edu.ua/> Department of Pediatrics №2 #) to the teacher who conducts a practical lesson. To complete the missed lesson, the student must answer questions of the topic writing or orally.

The procedure for admission to the discipline final control is giving by the Regulations on the organization of the educational process in National Pirogov Memorial Medical University (link <https://www.vnmdu.edu.ua/General> information / Basic documents). Students who do not have missed practical classes and lectures and received an average traditional grade of at least "3" are allowed to pass final control.

Additional points. Individual points in the discipline that student can receive for individual work, the amount of which is published on the website of the department in the teaching materials of the discipline and is determined by the results of his individual work according to Regulation of the Academic process in National Pirogov Memorial Medical University (link <https://www.vnmdu.edu.ua/General> information).

Conflict resolution. In case of misunderstandings and complaints to the teacher because of the quality of educational services, knowledge assessment and other conflict situations, student should submit his / her claims to the teacher. If the issue is not resolved, the student has a right to apply to the head of the department according to Complaints Consideration Procedure ([https://www.vnmdu.edu.ua/General information / Basic documents](https://www.vnmdu.edu.ua/General%20information%20-%20Basic%20documents)).

Politics in time of remote learning. Distance learning regulated by the Regulations of the elements of remote learning in National Pirogov Memorial Medical University ([https://www.vnmdu.edu.ua/ General information](https://www.vnmdu.edu.ua/General%20information)). The main training platforms for studying are Microsoft Team and Google Meets. Practical classes and lectures, exercises and consultations during distance learning is published on the website of the department ([https://www.vnmdu.edu.ua/ Department of Pediatrics№2/Student](https://www.vnmdu.edu.ua/Department%20of%20Pediatrics%20-%20Student) or [https://www.vnmdu.edu.ua/Department of Pediatrics№2/News](https://www.vnmdu.edu.ua/Department%20of%20Pediatrics%20-%20News)).

Feedback from teachers is via messengers (Viber, Telegram, WhatsApp) or e-mail (at the teacher's choice) during working hours.

8. Educational resources.

Educational and methodological support of the discipline is published on the website of the department ([https://www.vnmdu.edu.ua/ Department of Pediatrics№2/ To students](https://www.vnmdu.edu.ua/Department%20of%20Pediatrics%20-%20To%20students)).

1) LIST OF RECOMMENDED LITERATURE

1. Manolio TA, Collins FS, Cox NJ, Goldstein DB, Hindorff LA, Hunter DJ, et al. Finding the missing heritability of complex diseases. *Nature*. 2009;747–53. doi: 10.1038/nature08494.
2. McCarthy MI, Abecasis GR, Cardon LR, Goldstein DB, Little J, Ioannidis JP, et al. Genome-wide association studies for complex traits: consensus, uncertainty and challenges. *Nat Rev Genet*. 2008;9:356–69. doi: 10.1038/nrg2344.
3. Schork NJ, Murray SS, Frazer KA, Topol EJ. Common vs. rare allele hypotheses for complex diseases. *Curr Opin Genet Dev*. 2009;19:212–9. doi: 10.1016/j.gde.2009.04.010.
4. Bodmer W, Bonilla C. Common and rare variants in multifactorial susceptibility to common diseases. *Nat Genet*. 2008;40:695–701. doi: 10.1038/ng.f.136.
5. Goldstein DB. Common genetic variation and human traits. *N Engl J Med*. 2009;360:1696–8. doi: 10.1056/NEJMp0806284.
6. Zhu Q, Ge D, Maia JM, Zhu M, Petrovski S, Dickson SP, et al. A genome-wide comparison of the functional properties of rare and common genetic variants in humans. *Am J Hum Genet*. 2011;88:458–68. doi: 10.1016/j.ajhg.2011.03.008.
7. Lu YF, Goldstein DB, Angrist M, Cavalleri G. Personalized medicine and human genetic diversity. *Cold Spring Harb Perspect Med*. 2014;4:a008581. doi: 10.1101/cshperspect.a008581.
8. Crespi BJ. The emergence of human-evolutionary medical genomics. *Evol Appl*. 2011;4:292–314. doi: 10.1111/j.1752-4571.2010.00156.x.

9. Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, Handsaker RE, et al. An integrated map of genetic variation from 1,092 human genomes. *Nature*. 2012;**491**:56–65. doi: 10.1038/nature11632.
10. 1000 Genomes Project Consortium A global reference for human genetic variation. *Nature*. 2015;**26**:68–74.
11. Sousa V, Peischl S, Excoffier L. Impact of range expansions on current human genomic diversity. *Curr Opin Genet Dev*. 2014;**29**:22–30. doi: 10.1016/j.gde.2014.07.007.

2) Electronic resources:

1. University site <http://vnmue.edu.ua>
2. Library of University: <http://library.vnmue.edu.ua>
3. Test Center of Ministry of Health of Ukraine <https://www.testcentr.org.ua/uk/>
4. Ministry of Health of Ukraine <https://moz.gov.ua/>
5. Center of Public Health of Ministry of Health of Ukraine <https://phc.org.ua/kontrol-zakhvoryuvan>
6. Dr. Najeeb Lectures <https://www.youtube.com/channel/UCPHpx55tgrbm8FrYYCflAHw>
7. Osmosis <https://www.youtube.com/c/osmosis/>
8. MEDCRAM- Medical Lectures explained clearly <https://www.youtube.com/user/MEDCRAMvideos>
9. PubMed : <https://pubmed.ncbi.nlm.nih.gov/>


4) Methodical recommendations for practical classes and ISW, consultations (once a week according to the schedule of consultations).

9. The schedule and distribution of groups by teachers is published on the website of the department (<https://www.vnmue.edu.ua/> Department of Pediatrics №2 / Student).

10. Questions for the final control are published on the web page of the department (<https://www.vnmue.edu.ua/> Department of Pediatrics2 / Student).

The syllabus of the discipline «Pediatric Orphan disorders» was discussed and approved at the meeting of the department Pediatric №2 (record № 1, dated "30" "08" 2022)

Responsible for the academic discipline  associate professor of HEI Marina SHALAMAI

Head of the Department of Pediatrics №2  professor of HEI Veronika DUDNYK