



MINISTRY OF EDUCATION AND SCIENCE OF THE RUSSIAN  
FEDERATION  
Federal state autonomous educational institution  
of higher education  
«Far Eastern Federal University»  
(FEFU)

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**SCHOOL OF BIOMEDICINE**

«AGREED»

Head of education program  
«General medicine»

Khotimchenko Yu.S.

(signature)

(Full name)

«09» of July 2019

«APPROVED»

Director of the Department of Clinical  
Medicine

Geltser B.I.

(signature)

(Full name)

«09» of July 2019



**WORKING PROGRAM OF ACADEMIC DISCIPLINE (WPAD)**

« Medical genetics»

Educational program

Specialty 31.05.01 «General medicine»

**Form of study: full time**

year 4 semester 8

lectures 10 hours

practical classes not provided

laboratory works not provided

total amount of in-classroom work 10 hours

independent self-work 26 hours

control works ( )

credit 8 semester

exam not provided

The working program is drawn up in accordance with the requirements of the Federal state educational standard of higher education (level of training), approved by the order of the Ministry of education and science of the Russian Federation from 09.02.2016 № 95.

The working program of the discipline was discussed at the meeting of the Department of fundamental and clinical medicine. Protocol No. 8, 09 of July 2019

Author: prof.. Ovchinnicova A.A.

## Annotation

The discipline " Medical genetics" is intended for students enrolled in the educational program 31.05.01. "General Medicine", included in the basic part of the curriculum, is implemented in the 4<sup>th</sup> year in 8 semester. The total complexity of the discipline is 36 hours, 1 credits

In developing the work program of the discipline, the Federal State Educational Standard of Higher Education in the specialty 31.05.01 "General Medicine" (level of specialty), the curriculum for preparing students were used.

The course program is based on the basic knowledge gained by students: the ability to abstract thinking, analysis, synthesis (CC-1); the willingness to solve common tasks of professional activity with the use of information and bibliographic resources , biomedical terminology , information and communication technologies , taking into account the main requirements for information security (GPC – 1)

**The purpose of the** discipline is to master the theoretical foundations of the diagnosis, prevention and treatment of hereditary diseases by students. When studying the section of medical genetics, students will become familiar with the patterns of transmission in generations of hereditary diseases, the formation of multifactorial pathology involving genetic factors, aspects of the occurrence of sporadic congenital pathology, and de nova mutation.

**The main task** is to teach students a clinical approach to the assessment of hereditary pathology.

As a result of studying this discipline, students form the following general professional and professional competencies:

Competence code and formulation	Stages of forming the competence	
the readiness for medical use of drugs and other medical substances and their combinations in solving	Knows	Principles of etiological, pathogenetic, symptomatic treatment of the main hereditary diseases
	Is able to	To prescribe pathogenetic therapy taking into

professional problems (GPC – 8)		account the etiology of the disease with the use of drug therapy in patients who need medical rehabilitation.
	Possesses	Methods of providing medical care
the readiness to collect and to analyze patient complaints, data of its history, the results of laboratory, instrumental, postmortem and other examinations to recognize the incidence or the absence of diseases (PC – 5)	Knows	1. maintaining a typical accounting and reporting medical records in medical organizations 2. Fundamentals of preventive medicine, the organization of preventive measures aimed at improving the health of the population
	Is able to	1. plan, analyze and evaluate the quality of medical care, the state of public health and the influence of environmental and production factors on it 2. assess the social factors affecting the patient's physical and psychological health: cultural, ethnic, religious, individual, family, social risk factors; make a preliminary diagnosis - to synthesize information about the patient in order to determine the pathology and the causes of its cause; 3. to outline the amount of additional studies in accordance with the prognosis of the disease, to clarify the diagnosis and obtain a reliable result
	Possesses	1. Proper medical record keeping 2. Methods of general clinical examination
the ability of determining the patient's basic pathological conditions , symptoms, syndromes, diseases in accordance with the International Statistical Classification of Diseases and problems related to health , the 10th review. (PC – 6)	Knows	Principles of etiological, pathogenetic, symptomatic treatment of the main diseases of the central nervous system and peripheral nervous system. Providing emergency and emergency care, indications and contraindications for prescribing therapeutic measures, evaluation of treatment results
	Is able to	Assign pathogenetic therapy based on the etiology of the disease
	Possesses	Methods of providing medical care
the ability to determining the tactics of patient surveillance with different nosological entities. (PC – 8)	Knows	Principles of etiological, pathogenetic, symptomatic treatment of the main diseases of the central nervous system and peripheral nervous system. Providing emergency and emergency care, indications and

		contraindications for prescribing therapeutic measures, evaluation of treatment results
	Is able to	Assign pathogenetic therapy based on the etiology of the disease
	Possesses	Methods of providing medical care

## **I. STRUCTURE AND CONTENT OF THEORETICAL PART OF THE COURSE (10 HOURS)**

### **Section 1. MEDICAL GENETICS**

#### **Theme 1. Propaedeutics of hereditary pathology. Prevention of hereditary pathology (2 hours)**

Human genome. Classification of hereditary diseases. General clinical features of the manifestations of hereditary diseases. Principles and methods of diagnosis of hereditary diseases. Medical genetic counseling.

#### **Theme 2. Hereditary metabolic disorders (2 hours)**

Violations of amino acid metabolism (aminoacidopathy): types of inheritance, clinical characteristics, general principles of diagnosis. Phenylketonuria. Hereditary disorders of carbohydrate metabolism. Natural disorders of lipid metabolism. Mucopolysaccharidosis. Orphan Diseases (Pompe Disease, Fabry Disease)

#### **Theme 3 Hereditary neuromuscular diseases (2 hours)**

Primary muscular diseases (myopathies, hereditary muscular dystrophies) Hereditary polyneuropathy. Hereditary spinal amyotrophy

#### **Theme 4. Hereditary diseases of the extrapyramidal system (2 hours)**

Torsion dystonia. Chorea Goettington, Hepato-lenticular degeneration

#### **Theme 5. Hereditary Spinocerebellar Ataxia (2 hours)**

Neurofibromatosis (Reklingauzena disease). Ataxia - telangiectasia (Louis-Bar disease). Friedreich's Disease

## **THE STRUCTURE AND CONTENT OF THE PRACTICAL PART OF THE COURSE**

The educational and methodological support of self – study work in the discipline "Neurology, medical genetics" is presented in Supplement 1 and includes: a schedule for the implementation of independent work in the discipline, including approximate time limits for the implementation of each

task; characteristics of tasks for self – study work and methodological recommendations for their implementation; requirements for the presentation and presentation of the results of independent work; criteria for assessing the performance of independent work.

### CONTROL FOR ACHIEVEMENT OF THE COURSE GOALS

№	Controlled sections/topics of the discipline	Codes and stages of forming the competences		Means for evaluation	
				Current control	Half-way attestation
	<b>Section 1. Medical genetics</b>	GPC-8 the readiness for medical use of drugs and other medical substances and their combinations in solving professional problems	Knows	OS-1 Interview, Test WW-1, Abstract WW-7	OS-1 Interview, Test WW-1
			Is able to	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview, Test WW-1,
			Possesses	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview Case task WW-11
	<b>Section 1. Medical genetics</b>	PC – 5 the readiness to collect and to analyze patient complaints, data of its history, the results of laboratory, instrumental postmortem and other examinations in order to recognize the incidence or the absence of diseases	Knows	OS-1 Interview, Test WW-1, Abstract WW-7	OS-1 Interview, Test WW-1
			Is able to	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview, Test WW-1,
			Possesses	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview Case task WW-11
	<b>Section 1. Medical genetics</b>	PC-6 the ability of determining the	Knows	OS-1 Interview, Test WW-1, Abstract WW-7	OS-1 Interview, Test WW-1

		patient's basic pathological conditions, symptoms, syndromes, diseases in accordance with the International Statistical Classification of Diseases and problems related to health, the 10th review.	Is able to	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview, Test WW-1,
			Possesses	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview Case task WW-11
<b>Section 1. Medical genetics</b>	PC – 8 the ability to determining the tactics of patient surveillance with different nosological entities.		Knows	OS-1 Interview, Test WW-1, Abstract WW-7	OS-1 Interview, Test WW-1
			Is able to	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview, Test WW-1,
			Possesses	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview Case task WW-11

Control and methodological materials, as well as criteria and indicators necessary for the assessment of knowledge and skills and characterizing the stages of the competencies formation are presented in Supplement 1.

## **II. LIST OF EDUCATIONAL LITERATURE AND INFORMATIONAL-METHODICAL REQUIREMENTS FOR THE DISCIPLINE.**

### **Main literature**

*(electronic and print publications)*

### **Additional literature**

1. Medical Ethics in Clinical Practice / Springer Nature Switzerland AG 2016  
<https://link.springer.com/book/10.1007/978-3-030-00719-5#authorsandaffiliationsbook>
2. Clinical Relevance of Genetic Factors in Pulmonary Diseases / Springer Nature Singapore Pte Ltd. 2018  
<https://link.springer.com/book/10.1007/978-981-10-8144-6#editorsandaffiliations>
3. Rare Diseases Epidemiology: Update and Overview / Springer International Publishing AG 2017  
<https://link.springer.com/book/10.1007/978-3-319-67144-4#editorsandaffiliations>

## LIST OF INFORMATION TECHNOLOGIES AND SOFTWARE

The location of the computer equipment on which the software is installed, the number of jobs	List of licensed software
Multimedia auditorium Vladivostok Russian island, Ayaks 10, building 25.1, RM. M723 Area of 80.3 m2 (Room for independent work)	Windows Seven enterprice SP3x64 Operating System Microsoft Office Professional Plus 2010 office suite that includes software for working with various types of documents (texts, spreadsheets, databases, etc.); 7Zip 9.20 - free file archiver with a high degree of data compression; ABBYY FineReader 11 - a program for optical character recognition; Adobe Acrobat XI Pro 11.0.00 - software package for creating and viewing electronic publications in PDF; WinDjView 2.0.2 - a program for recognizing and viewing files with the same format DJV and DjVu.

In order to provide special conditions for the education of persons with disabilities all buildings are equipped with ramps, elevators, lifts, specialized places equipped with toilet rooms, information and navigation support signs



#### **4. METHODOLOGICAL RECOMMENDATIONS ON THE COMPLETING THE DISCIPLINE**

The main source of information and the knowledge-forming component of the discipline " Neurology, Medical Genetics" is a series of lectures.

*Students guidelines:*

1. Students must attend all the lectures and note-taking the material presented.
2. The assimilation and consolidation of lecture materials should be carried out in first days after listening to a lecture.
3. First, it is necessary to study the lecture notes, diagrams and figures. If necessary, read to the recommended literature.
4. In conclusion, try to answer the questions of the lecture plan.
5. In case of missing a lecture, study the material on the lecture topic using the recommended literature. This significantly increases self-preparation time.
6. It is necessary to return to the lecture materials again: while preparing for the final lesson; in preparation for the final control (it is necessary to pay attention to the control questions).

The main goal of the discipline is formation and development skills and ability to assess the physiological state of the nervous system, to interpret data of laboratory, clinical and functional diagnostics for the solution of professional tasks. At the end of the course the student should have the skills for diagnosis the major pathological states, symptoms, syndromes, diseases, nosological forms. Students are encouraged to systematically study the teaching material using textbooks, textx and methodical writings in accordance with the study plan, and to perform all task in a timely manner, which is especially important when using grade-rating system for assessing students' knowledge.

The goal of students' individual work is to master fundamental knowledge, professional skills and experiences of their specialty, experience of creative scientific research. Individual work of students promotes the development of autonomy, responsibility and organization, creative approach to solving the problems of the educational and professional level, deepen and broaden knowledge, formation of interest to cognitive activity, mastering the techniques of learning, the development of cognitive abilities.

Individual work of students for the discipline Neurology, medical genetics is mandatory for each student, its volume is determined by the federal educational

standard and curriculum. It is necessary at the very beginning of the course to carefully plan the time allocated for individual work with the sources and literature on the subject.

Individual work includes:

- a) reading textbooks, lectures, methodical recommendations, scientific articles
- b) reading and analyzing literature passages of journalistic nature;
- c) reading and analysis of literary passages of scientific nature;
- g) working with resources posted on the Internet.

The purpose of this types of work is to instill an interest in reading and to teach students to overcome difficulties in reading, extract the necessary information from the text to teach them to use Russian and International sources for self-education and improve their professional skills

#### **IV. MATERIAL AND TECHNICAL SUPPORT OF DISCIPLINE**

For practical work, as well as for the organization of independent work, students have access to the following laboratory equipment and specialized classrooms that meet applicable sanitary and fire regulations, as well as safety requirements for educational and research and production work:

<b>Name of equipped premises and rooms for independent work</b>	<b>List of basic equipment</b>
690922, Primorsky Krai, Vladivostok, island Russian, the Saperny Peninsula, the village of ayaks, 10, RM. M 421	Multimedia audience: Monoblock Lenovo C360G-i34164G500UDK; projection Screen Projecta Elpro Electrol, 300x173 cm; Multimedia projector, Mitsubishi FD630U, 4000 ANSI Lumen 1920 x 1080; Flush interface with automatic retracting cables TLS TAM 201 Stan; Avervision CP355AF; lavalier Microphone system UHF band Sennheiser EW 122 G3 composed of a wireless microphone and receiver; Codec of videoconferencing LifeSizeExpress 220 - Codeonly - Non-AES; Network camera Multipix MP-HD718; Two LCD panel, 47", Full HD, LG M4716CCBA; Subsystem of audiocommentary and sound reinforcement; centralized uninterrupted power supply



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**SCHOOL OF BIOMEDICINE**

**TRAINING AND METHODOLOGICAL SUPPORT**

**INDEPENDENT WORK OF STUDENTS**

Medical Genetics

Specialty 31.05.01. General Medicine

**Form of training full-time**

**Vladivostok**

**2018**

### Schedule of the independent work on the discipline

№	Type of self-study work	Self-study work	Estimated time to run	Form of control
1.	Lecture preparation	Preparation for lectures based on educational material and work with literature Preparing for the test based on knowledge about the mechanisms of heredity.	16 hr.	VR-1 Interview YO-2 Colloquium WW-1 Test WW -7 Abstract WW -11 Case task
2	Individual preparation	Consolidation of the material studied (work with lecture materials, educational literature);	10 hr.	VR-1 Interview WW -7 Abstract WW -11 Case task



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**SCHOOL OF BIOMEDICINE**

**FUND OF ASSESSMENT TOOLS**

Medical Genetics

Educational program

Specialty 31.05.01. General Medicine

**Form of training full-time**

Vladivostok  
2018

## Passport of assessment fund

Completed in accordance with the Regulations on the Funds of Evaluation Assets of Educational Programs of Higher Education - Bachelor's Programs, Specialties, FEFU Magistrates, approved by order of the Rector No. 12-13-850 of May 12, 2015.

Competence code and formulation	Stages of forming the competence	
GPC – 8 the readiness for medical use of drugs and other medical substances and their combinations in solving professional problems	Knows	Principles of etiological, pathogenetic, symptomatic treatment of the main hereditary diseases
	Is able to	To prescribe pathogenetic therapy taking into account the etiology of the disease with the use of drug therapy in patients who need medical rehabilitation.
	Possesses	Methods of providing medical care
PC – 5 the readiness to collect and to analyze patient complaints, data of its history, the results of laboratory, instrumental postmortem and other examinations in order to recognize the incidence or the absence of diseases	Knows	<ol style="list-style-type: none"> <li>1. maintaining a typical accounting and reporting medical records in medical organizations</li> <li>2. Fundamentals of preventive medicine, the organization of preventive measures aimed at improving the health of the population</li> </ol>
	Is able to	<ol style="list-style-type: none"> <li>1. plan, analyze and evaluate the quality of medical care, the state of public health and the influence of environmental and production factors on it</li> <li>2. assess the social factors affecting the patient's physical and psychological health: cultural, ethnic, religious, individual, family, social risk factors; make a preliminary diagnosis - to synthesize information about the patient in order to determine the pathology and the causes of its cause;</li> <li>3. to outline the amount of additional studies in accordance with the prognosis of the disease, to clarify the diagnosis and obtain a reliable result</li> </ol>
	Possesses	<ol style="list-style-type: none"> <li>1. Proper medical record keeping</li> <li>2. Methods of general clinical examination</li> </ol>
PC – 6 the ability of determining the patient's basic pathological conditions,	Knows	Principles of etiological, pathogenetic, symptomatic treatment of the main diseases of the central nervous system and peripheral nervous system. Providing emergency and emergency care, indications and

symptoms, syndromes, diseases in accordance with the International Statistical Classification of Diseases and problems related to health, the 10th review.		contraindications for prescribing therapeutic measures, evaluation of treatment results
	Is able to	Assign pathogenetic therapy based on the etiology of the disease
	Possesses	Methods of providing medical care
PC – 8 the ability to determining the tactics of patient surveillance with different nosological entities.	Knows	Principles of etiological, pathogenetic, symptomatic treatment of the main diseases of the central nervous system and peripheral nervous system. Providing emergency and emergency care, indications and contraindications for prescribing therapeutic measures, evaluation of treatment results
	Is able to	Assign pathogenetic therapy based on the etiology of the disease
	Possesses	Methods of providing medical care

### CONTROL FOR ACHIEVEMENT OF THE COURSE GOALS

№	Controlled sections/topics of the discipline	Codes and stages of forming the competences		Means for evaluation	
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	<b>Section 1. Medical genetics</b>	GPC-8 the readiness for medical use of drugs and other medical substances and their combinations in solving professional problems	Knows	OS-1 Interview, Test WW-1, Abstract WW-7	OS-1 Interview, Test WW-1
			Is able to	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview, Test WW-1,
			Possesses	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview Case task WW-11
	<b>Section 1. Medical genetics</b>	PC – 5 the readiness to collect and to	Knows	OS-1 Interview, Test WW-1, Abstract WW-7	OS-1 Interview, Test WW-1

		analyze patient complaints, data of its history, the results of laboratory, instrumental postmortem and other examinations in order to recognize the incidence or the absence of diseases	Is able to	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview, Test WW-1,
			Possesses	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview Case task WW-11
	<b>Section 1. Medical genetics</b>	PC-6 the ability of determining the patient's basic pathological conditions, symptoms, syndromes, diseases in accordance with the International Statistical Classification of Diseases and problems related to health, the 10th review.	Knows	OS-1 Interview, Test WW-1, Abstract WW-7	OS-1 Interview, Test WW-1
			Is able to	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview, Test WW-1,
			Possesses	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview Case task WW-11
	<b>Section 1. Medical genetics</b>	PC – 8 the ability to determining the tactics of patient surveillance with different nosological entities.	Knows	OS-1 Interview, Test WW-1, Abstract WW-7	OS-1 Interview, Test WW-1
			Is able to	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview, Test WW-1,
			Possesses	OS-1 Interview Case task WW-11 Medical history case	OS-1 Interview Case task WW-11



## Scale of assessment of the level of competence formation

Competence code and formulation	Stages of competence formation		Criteria	Indicators	Score
GPC-8 the readiness for medical use of drugs and other medical substances and their combinations in solving professional problems	Knows (the threshold level)	Principles of etiological, pathogenetic, symptomatic treatment of major diseases of the central nervous system and peripheral nervous system.	Knowledge of etiological principles, pathogenetic, symptomatic treatment of major diseases of the central nervous system and peripheral nervous system.	The capacity and willingness to learn etiological principles of, pathogenetic, symptomatic treatment of major diseases of the central nervous system and peripheral nervous system.	65-71
	Is able to	Prescribe pathogenetic therapy taking into account the etiology of the disease with the use of drug therapy in patients in need of medical rehabilitation.	Knowledge of pathogenetic therapy taking into account the etiology of the disease with the use of drug therapy in patients in need of medical rehabilitation.	The capacity and willingness to learn principles of pathogenetic therapy taking into account the etiology of the disease with the use of drug therapy in patients in need of medical rehabilitation.	71-84
	Possesses	Methods of providing medical care	Skill of the correct rendering the medical help	Able to solve problems on the choice of methods of providing medical care	85-100
(PC – 5) the readiness to collect and to analyze patient complaints, data of its history, the results of laboratory, instrumental postmortem and other examinations in order to recognize the incidence or the absence of diseases.	Knows (threshold level)	1. Maintaining a typical accounting and reporting medical records in medical organizations 2. The basics of preventive medicine, the organization of preventive measures aimed at improving the health of the	1. principles of maintaining a standard accounting and reporting medical records in medical organizations 2. Fundamentals of preventive medicine, the organization of preventive measures aimed at	Formed and structured knowledge on the conduct of typical accounting and reporting medical documentation in medical organizations 2. Basic of preventive medicine, organization of prophylactic	65-71

		population	improving the health of the population	events aimed at improving the health of the population	
	Is able to (Advanced)	<p>1. plan, analyze and evaluate the quality of medical care, health status of the population and the influence of environmental and production factors on it</p> <p>2. to assess the social factors affecting the state of the physical and psychological health of the patient: cultural, ethnic, religious, individual, family, social risk factors; make a preliminary diagnosis - to synthesize information about the patient in order to determine the pathology and the causes of its callers;</p> <p>3. to outline the amount of additional research in accordance with the prognosis of the disease, to clarify the diagnosis and obtain a reliable result</p>	<p>The ability to plan, analyze and evaluate the quality of medical care, the state of health of the population and the impact on it of factors of the surrounding and industrial environment</p> <p>2. to assess the social factors affecting the state of physical and psychological health of the patient: cultural, ethnic, religious, individual, family, social risk factors; to make a preliminary diagnosis - to synthesize information about the patient in order to determine the pathology and the reasons for it;</p> <p>3. to outline the volume of additional studies in accordance with the prognosis of the disease, to clarify the diagnosis and obtain a reliable result</p>	<p>Ready and able to plan, analyze and evaluate the quality of medical care, the state of health of the population and the influence of environmental and production factors on it</p> <p>2. to assess the social factors influencing the state of the patient's physical and psychological health: cultural, ethnic, religious, individual, family, social risk factors; make a preliminary diagnosis - to synthesize information about the patient in order to determine the pathology and the reasons for it;</p> <p>3. to outline the volume of additional studies in accordance with the prognosis of the disease, to clarify the diagnosis and obtain a reliable result</p>	71-84
	Possesses (High level)	<p>1. The correct management of medical records</p> <p>2. Methods of general clinical examination</p>	<p>Skill 1. The correct management of medical documentation</p> <p>2. carrying out general clinical examination</p>	<p>Able to solve problems by choosing methods of general clinical examination.</p>	85-100

<p>PC-6 ability to determining the patients basic pathological conditions, symptoms, syndromes, diseases, clinical entities, in accordance with the International Statistical Classification of Diseases and Related Health X review</p>	<p>Knows (threshold level)</p>	<p>The physiological signs of major pathological conditions, symptoms, syndromes, diseases, clinical entities, in accordance with the International Statistical Classification of Diseases and Related Health X review</p>	<p>The main physiological signs of major pathological conditions, symptoms, syndromes, diseases, clinical entities, in accordance with the International Statistical Classification of Diseases and Related Health X review</p>	<p>The capacity and willingness to learn a foreign language at the level of everyday communication, to the written and oral communication in the official language</p>	<p>65-71</p>
	<p>To be able (Advanced)</p>	<p>Assign pathogenetic therapy based on the etiology of the disease</p>	<p>Ability to prescribe etio-patho-genetic therapy</p>	<p>Ready and able to prescribe pathogenetic therapy based on the etiology of the disease</p>	<p>71-84</p>
	<p>Possesses (High level)</p>	<p>Methods of providing medical care</p>	<p>The skill of providing medical care</p>	<p>Able to solve problems on the choice of methods of providing medical care</p>	<p>85-100</p>
<p>PC – 8 the ability to determining the tactics of patient surveillance with different nosological entities.</p>	<p>Knows (threshold level)</p>	<p>Principles of etiological, pathogenetic, symptomatic treatment of the main diseases of the central nervous system and peripheral nervous system. Principles of application of natural therapeutic factors, drug, non-drug therapy and other methods in patients in need of medical rehabilitation and sanatorium-resort treatment</p>	<p>Knowledge of the principles of etiological, pathogenetic, symptomatic treatment of the main diseases of the central nervous system and the peripheral nervous system. Principles of application of natural therapeutic factors, drug, non-drug therapy and other methods in patients in need of medical rehabilitation and sanatorium-resort treatment</p>	<p>Formed and structured knowledge of the principles of the etiological, pathogenetic, and symptomatic treatment of the main diseases of the CNS and the peripheral nervous system. Principles of the use of natural therapeutic factors, drug, non-drug therapy and other methods in patients in need of medical rehabilitation and spa-and-spa treatment</p>	<p>65-71</p>

	Is able to (advanced level)	To prescribe pathogenetic therapy taking into account the etiology of the disease using natural therapeutic factors, medicinal, non-drug therapy and other methods in patients who need medical rehabilitation and sanatorium-resort treatment	Ability to prescribe pathogenetic therapy taking into account the etiology of the disease with the use of natural therapeutic factors, medicinal, non-drug therapy and other methods in patients who need medical rehabilitation and sanatorium treatment	Ready and able to prescribe pathogenetic therapy taking into account the etiology of the disease with the use of natural therapeutic factors, drug, non-drug therapy and other methods in patients who need medical rehabilitation Spa treatment	71-84
	Possesses (High level)	Methods of providing medical care	Selection skill methods of providing medical care	Able to solve problems on the choice of methods of providing medical care	85-100

## **Guidelines that determine the results of the discipline evaluation procedures development**

Intermediate certification for the discipline "Neurology, medical genetics" is carried out in the form of offset. Offset includes 2 stages:

Stage 1 - written testing (conducted at the last lesson of the discipline cycle);

Stage 2 - final oral interview (includes three theoretical questions).

*Criteria for evaluating the test task:*

The mark "excellent" is given to a student, if the number of correct answers is 100%;

The mark "good" is given to a student, if the number of correct answers ranges from 81 to 99%;

The mark "satisfactory" is given to a student if the number of correct answers ranges from 65-70% to 80%;

A grade of "unsatisfactory" is given to a student if the number of correct answers is less than 65%;

*Evaluation criteria for an oral examination:*

The mark "excellent" is given to a student, if the answer to the question is complete, detailed, the student uses the basic teaching literature and lecture material, the student's oral speech is built logically, reasonably and clearly;

The mark "good" is given to the student, if the answer to the question is not sufficiently complete, the student uses the main academic literature;

The mark "satisfactory" is given to the student, if the answer to the question is fragmentary, the main educational literature is used poorly;

The rating "unsatisfactory" is given to the student, if the answer to the question is not received.

Integral assessment of the discipline is the arithmetic average of all stages of the exam.

"Excellent" deserves a student who has discovered systematic and deep knowledge of anatomy, physiology of the nervous system, questions of topical diagnostics, who can independently properly use acquired practical skills in examining a neurological patient, possessing full knowledge of the clinic, diagnosis and treatment of nervous diseases and emergency conditions for them specific curriculum.

"Well" is exposed to students who have shown the systematic nature of knowledge in a discipline, have insignificant gaps, do not affect the correct nature of the answer and are capable of self-replenishing the justification in the course of further study and professional activity.

"Satisfactorily" exhibit students who have committed errors on issues of general neurology, but who have the necessary knowledge to eliminate these errors

"Unsatisfactory" is exposed to students who have made gross errors in the answers and have significant gaps in knowledge

### **Evaluation tools for intermediate certification**

#### ***Full questions list for the test:***

1. Classification of hereditary diseases of the nervous system.
2. Classification of hereditary neuromuscular diseases.
3. Chorea Huntington. Clinic. Principles of therapy.
4. Spinocerebellar degeneration. Friedreich disease. Clinic. Medical genetic counseling.
5. Neurofibromatosis Reklingauzen. Clinic. Medical genetic counseling.
6. X-linked progressive muscular dystrophy (Duschen disease, Becker disease). Clinic. Medical genetic counseling.
7. Hereditary polyneuropathy Sharko-Marie disease) Clinic. Medical genetic counseling
8. Hereditary spinal amyotrophies (Verdnig-Hoffman's disease, Kugelberg's disease Velandar) Clinic. Medical genetic counseling
9. Hepatocerebral degeneration. Etiology, Pathogenesis, Clinic. Diagnostics. Treatment
10. Ataxy - Telangiectasia - Louis-Bar disease. Etiology, Pathogenesis, Clinic. Diagnostics. Treatment
11. Hereditary metabolic disorders of amino acids. Phenylketonuria. Etiology, pathogenesis, type of inheritance. Diagnosis, principles of therapy. Maternal Phenylketonuria.
12. Myasthenia. Etiology. Clinic. Diagnostics. Treatment.
13. Syringomyelia and syringomyelobulia. Clinic. Diagnostics. Treatment.
14. Hereditary disorders of lipid metabolism - pathogenesis, clinic, diagnosis, treatment.
15. Orphan diseases - Pompe disease. Etiology. Pathogenesis, clinic, diagnosis, treatment
16. Fabry disease. Etiology, Pathogenesis, clinic, diagnosis. Treatment
17. Clinical and paraclinical methods for the diagnosis of congenital and hereditary diseases.
18. Medical genetic counseling.

***Examples of test tasks:***

An organism is called homozygous, in somatic cells of which:

- a) different alleles
- b) one allele
- c) identical alleles
- d) there are no alleles

The number of alleles of a single gene in a mature germ cell of a diploid organism:

- a) one
- b) two
- c) at three o'clock
- d) four

For the diagnosis of monogenic diseases, the following methods are used:

- a) study of sex chromatin
- b) biochemical
- c) functional diagnostics
- d) cytogenetic

***Criteria for the test task evaluation:***

The mark “excellent” is given to the student, if the number of correct answers is 100%;

The mark “good” is given to the student, if the number of correct answers ranges from 81 to 99%;

The mark “satisfactory” is given to the student, if the number of correct answers is between 65-70% and 80%; the mark “unsatisfactory” is given to the student, if the number of correct answers is less than 65%;

Examples of questions for the oral survey on the topic "Epilepsy and paroxysmal states."

1. List the types of simple focal epileptic seizures.
2. What are the classification criteria for forms of epilepsy.
3. What are the main characteristics of an epileptic seizure?

Evaluation criteria for an oral response

The mark “excellent” is given to the student, if the answer to the question is complete, detailed, the student uses the basic teaching literature and lecture material, the student’s oral speech is built logically, reasonably and clearly;

The mark “good” is given to the student, if the answer to the question is not sufficiently complete, the student uses the main academic literature;

The mark “satisfactory” is given to the student, if the answer to the question is fragmentary, the main educational literature is used poorly;

The mark “unsatisfactory” is given to the student, if the answer to the question is not received;

Examples of situational tasks on the topic "Tumors and abscesses of the brain"

A child of 10 years old for 2 months is bothered by headaches that occur mostly in the morning after sleep. Today, the child has a seizure with loss of consciousness and generalized tonic-clonic convulsions, which began with the tonic tension of the left hand. On examination, a decrease in strength in the left hand to 4 points, an increase in tendon reflexes in the left extremities, a symptom of Babinsky on the left, were revealed.

1. Highlight pathological syndromes.

2. Put a topical and presumptive clinical diagnosis. 3. Make a survey plan.

Answer: 1. Left-sided central hemiparesis, cerebral syndrome, somato-motor, secondary-generalized epileptic seizure.

2. The right frontal lobe in the area of the middle parts of the precentral gyrus is affected. A brain tumor.

3. CT scan or MRI of the brain.

Criteria for assessing the situational problem:

The mark “excellent” is given to the student, if the answer is complete, detailed, the student clearly answered all the points of the problem questions, the answer is built logically, correctly, clearly argued;

The mark “good” is given to the student, if the answer to the question is not complete enough, the student answered only part of the questions of the problem.

The mark “satisfactory” is given to the student, if the answer to the question is fragmentary, the main educational literature is used poorly;

The mark “unsatisfactory” is given to the student, if the answer to the question is not received;