Appendix to the working program

Federal State Budgetary Educational Institution of Higher Education
"Privolzhsky Research Medical University"
Ministry of Health of the Russian Federation

BANK OF ASSESSMENT TOOLS FOR DISCIPLINE

BASICS OF CLINICAL GENETICS

Training program (specialty): **31.05.01 GENERAL MEDICINE**

Department: **HOSPITAL PEDIATRICS**

Mode of study **FULL-TIME**

Nizhniy Novgorod
2023

1. Bank of assessment tools for the current monitoring of academic performance, mid-term assessment of students in the discipline

This Bank of Assessment Tools (BAT) for the discipline `Basics of clinical genetics` is an integral appendix to the working program of the discipline `Basics of clinical genetics`. All the details of the approval submitted in the WPD for this discipline apply to this BAT.

(Banks of assessment tools allow us to evaluate the achievement of the planned results stated in the educational program.

Assessment tools are a bank of control tasks, as well as a description of forms and procedures designed to determine the quality of mastering study material by students.)

2. List of assessment tools

The following assessment tools are used to determine the quality of mastering the academic material by students in the discipline/ practice:

No.	Assessment tool	Brief description of the assessment tool	Presentation of the assessment tool in the BAT
1	Tests	A system of standardized tasks that allows you to automate the procedure of measuring the level of knowledge and skills of a student	Bank of test tasks
2	Interview	A tool of control organized as a special conversation between the teacher and the student on topics related to the discipline being studied, and designed to clarify the amount of knowledge of the student on a specific section, topic, problem, etc.	Questions on topics/sections of the discipline

3. A list of competencies indicating the stages of their formation in the process of mastering the educational program and the types of evaluation tools

Code and formulation of competence*	Stage of competence formation	Controlled sections of the discipline	Assessment tools
UC-1 OPC-4 OPC-7 OPC-10 PC-5 PC-6 PC-7	Current	Section 1. Introduction to clinical genetics. Personalized diagnosis and treatment of hereditary diseases. Medical genetic counseling.	test tasks, Interview
UC-1 OPC-4 OPC-7 OPC-10 PC-5 PC-6 PC-7	Current	Section 2. Modern genetic research methods.	test tasks, Interview

UC-1 OPC-4 OPC-7 OPC-10 PC-5 PC-6 PC-7	Current	Section 3. Chromosomal syndromes. Prenatal diagnosis of congenital and hereditary diseases.	test tasks, Interview
UC-1 OPC-4 OPC-7 OPC-10 PC-5 PC-6 PC-7	Mid-term	Section 1. Introduction to clinical genetics. Personalized diagnosis and treatment of hereditary diseases. Section 2. Modern genetic research methods. Medical genetic counseling. Section 3. Chromosomal syndromes. Prenatal diagnosis of congenital and hereditary diseases.	Test tasks

4. The content of the assessment tools of entry, current control

4. The content of the evaluation means of current control

Current control is carried out by the teacher of the discipline when conducting classes in the form of: testing, interview.

4.1. Test tasks for assessing competencies: UC-1, GPC-4, GPC-7, GPC-10, PC-5, PC-6, PC-7.

Test tasks	Competence code (according to RPD)
1. PROBAND IS 1) Patient visiting a doctor 2) A healthy person who applied to a medical genetic consultation 3) A person who first came under the supervision of a geneticist 4) The person from whom the collection of a pedigree begins	UC-1 OPC-4 OPC-7 OPC-10 PC-5 PC-6 PC-7
2. SIBS IS 1) All relatives of the proband 2) proband's uncle 3) Proband's parents 4) Brothers and sisters of the proband	
3. THE OBJECT OF STUDY OF CLINICAL GENETICS ARE 1) A sick man 2) Sick and sick relatives 3) The patient and all members of his family, including healthy	
4. WHAT IS THE PROBABILITY OF THE BIRTH OF A SICK CHILD BY A WOMAN HAVING A SICK SON AND BROTHER WITH HEMOPHILIA 1) 25% 2) 50% 3) 100% 4) close to 0%	
5. DOLICHOCEPHALY IS	

1) Long narrow skull with protruding forehead and occiput 2) An increase in the longitudinal size of the skull relative to the transverse 3) An increase in the transverse size of the skull with a relative decrease in the longitudinal size 4) Expansion of the skull in the occipital and narrowing in the frontal part	
6. EPIKANT IS 1) unibrow 2) Widely spaced eyes 3) Vertical skin fold at the inner corner of the eye 4) Narrowing of the palpebral fissure	
7. OLIGODACTYLY IS 1) Lack of fingers 2) Finger fusion 3) Missing one or more fingers 4) Increasing the number of fingers	
8. CRYPTORCHISM IS 1) urinary tract infection 2) Undescended testicles into the scrotum 3) Underdevelopment of the genital organs	
9. ARACCHNODACTYLY IS 1) Finger shortening 2) Changing the shape of the fingers 3) Increasing the length of the fingers	
10. SYNDACTYLY IS 1) Fusion of limbs along the entire length 2) Limb fusion in the lower third 3) Finger fusion	
11. BRACHYCEPHALY IS 1) Expansion of the skull in the occipital and narrowing in the frontal part 2) "tower skull" 3) An increase in the transverse size of the head with a relative decrease in the longitudinal size 4) An increase in the longitudinal size of the skull relative to the transverse	
12. ANOPHTHALMIA IS 1) Congenital absence of eyeballs 2) congenital absence of the iris 3) Reduced distance between the inner corners of the eye sockets	
13. MICROGNATIA IS 1) Small size of the lower jaw 2) Small size of the upper jaw 3) small mouth opening	
14. HETEROCROMIA OF THE IRIS IS	

1) Abnormal color perception
2) Different coloration of the iris
3) Differences in the size of the irises

15. THE MAIN OBJECTIVES OF MEDICAL GENETICS IS THE STUDY

1) laws of heredity and variability of the human body
2) population statistics of hereditary diseases
3) molecular and biochemical aspects of heredity
4) changes in heredity under the influence of environmental factors
5) all of the above

16. THE PERINATAL PERIOD IS THE PERIOD WHICH

1) begins at 22 completed weeks of gestation and ends 7 completed days after birth.
2) begins at 28 completed weeks of gestation and ends 7 completed days after birth.
3) begins at 28 completed weeks of gestation and ends 28 completed days after birth.
4) begins at 22 completed weeks of gestation and ends 28 completed days after birth.

17. WHAT IS THE CRITICAL PERIOD OF DEVELOPMENT

1) this is a period of development characterized by increased sensitivity of the embryo and the embryo to the damaging effects of various factors
2) this is the period of formation of the functional system mother-placenta-fetus
3) this is the embryonic period
4) it's a blast period

18. TERATOGENIC TERMINATION PERIOD -

1) This is a period of intensive histogenesis and organogenesis
2) this is the period during which damaging factors cause a specific malformation.
3) this is the 15-40th days of intrauterine life
4) these are the first 2 weeks after conception, including the first 5-7 days of implantation.

19. INDICATIONS FOR MANDATORY PERICONCEPTIONAL PREVENTION OF CM

1) Absence of antenatal risk factors
2) Pregnancy with a second and subsequent child
3) Genetic risk of multifactorial congenital malformations
4) Young and healthy parents

20. PERICONCEPTIVE PREVENTION INCLUDES

1) consultation of a geneticist and other specialists before pregnancy, ultrasound at the recommended time of pregnancy, screening of serum markers, invasive diagnostic methods (if indicated).
2) consultation with a geneticist and other specialists before pregnancy
3) Ultrasound at the recommended time of pregnancy, screening of serum markers, invasive diagnostic methods (according to indications).
4) observation of a woman during pregnancy.

21. CHROMOSOMAL DISEASES ARE

1) all conditions characterized by disturbances in the structure of chromosomes
2) all conditions characterized by abnormalities in the number of chromosomes
3) all conditions characterized by disturbances in the structure or number of chromosomes
4) all conditions characterized by gene mutations.

22. THE COMBINATION OF ULTRASOUND AND BIOCHEMICAL RESEARCH IN TIME IS RECOGNIZED AS THE MOST EFFECTIVE

1) 11 weeks to 13 weeks
2) from 11 weeks to 13 weeks 6 days
3) 11 weeks to 12 weeks 6 days
4) from 10 weeks to 13 weeks 6 days

23. FOR FRUIT WITH CHROMOSOME DISEASES IS CHARACTERISTIC

1) reduction in the thickness of the collar space less than 2.5 mm
2) disappearance of the collar space after 14 weeks of pregnancy
3) increase in the thickness of the collar space more than 2.5 mm
4) a directly proportional relationship between a decrease in the thickness of the collar space and the risk of developing chromosomal diseases

24. REDUCING THE LEVEL OF PAPP-A INDICATES ABOUT

1) large intrauterine baby with developmental advance compared to normal gestational age
2) risk of fetal chromosomal abnormalities
3) intrauterine growth retardation
4) the need for invasive methods of prenatal diagnosis

25. THE MOST WIDELY INVASIVE PRENATAL DIAGNOSIS IS USED FOR

1) exclusion of congenital malformations
2) fetal sex determination
3) correction of pregnancy management
4) exclusion of fetal chromosomal disorders

test item number	response standard number	test item number	response standard number
1.	4	21.	3
2.	4	22.	2
3.	3	23.	3
4.	2	24.	2
5.	1	25.	4
6.	3		
7.	3		
8.	2		
9.	3		

10.	3		
eleven.	1		
12.	1		
13.	1		
14.	2		
15.	3		
16.	1		
17.	1		
18.	2		
19.	3		
20.	1		

4.2. Interview questions for assessing competencies: UC-1, GPC-4, GPC-7, GPC-10, PC-5, PC-6, PC-7.

1. The place of hereditary pathology in the structure of human diseases.
2. The role of hereditary and environmental factors in the pathogenesis of diseases.
3. Modern methods of diagnostics of hereditary diseases.
4. genealogical method.
5. Tactics of managing a patient with hereditary diseases.
6. neonatal screening.
7. Medical genetic counseling: tasks and indications for implementation.
8. Medical genetic counseling: principles and stages of counseling.
9. Factors causing human chromosomal diseases.
10. Characteristics of the main human chromosomal diseases.
11. Modern methods of diagnosing chromosomal diseases.
12. Modern methods of treatment and rehabilitation of patients with chromosomal diseases.
13. Prevention of chromosomal diseases.
14. Prenatal diagnosis of congenital and hereditary diseases.

5. Contents of evaluation tools for intermediate certification

Intermediate certification is carried out in the form of final testing.

5.1 The list of control tasks and other materials necessary for assessing knowledge, skills and experience: tests for sections of the discipline: test tasks.

5.1.1. Test questions with answer options for the test in the discipline "Basics of Clinical Genetics".

Test tasks	Competence code (according to WPD)
1. THE PRESENCE IN ONE PERSON OF MULTIPLE VARIANTS OF THE CHROMOSOMAL SET IS CALLED 1) Chromism 2) polyploidy 3) genetic cargo 4) mosaicism	UC-1 OPC-4 OPC-7 OPC-10 PC-5 PC-6 PC-7
2. PHENOTYPICAL SIGNS OF CHROMOSOMAL DISEASES ARE 1) mental development disorders 2) developmental disorders 3) multiple malformations 4) all of the above	
3. RECESSIVE TYPE OF INHERITANCE ASSOCIATED WITH	

THE X-CHROMOSOME IS CHARACTERIZED IN THAT

1) the ratio of affected males in each generation is 2:1
2) only men get sick
3) only women get sick
4) signs of the disease must be found in the mother of the proband

4. DOMINANT GENE IS THIS GENE, THE ACTION OF WHICH

1) found in a heterozygous state
2) found in the homozygous state
3) detected in hetero- and homozygous state
4) all of the above is incorrect

5. PHENOTYPE IS A SET OF FEATURES AND PROPERTIES OF THE ORGANISM, THE MANIFESTATION OF WHICH IS DUE TO

1) the action of a dominant gene
2) the action of a recessive gene
3) both dominant and recessive genes
4) interaction of the genotype with environmental factors

6. KARYOTYPE IS A SET OF FEATURES OF THE CELL CHROMOSOMAL SET, DETERMINED

1) number of sex chromosomes
2) shape of chromosomes
3) chromosome structure
4) all of the above
5) none of the above

7. AUTOSODOMINANT INHERITANCE IS DIFFERENT

1) predominantly affecting males
2) predominance in the generation of sick family members
3) manifestation of a pathological inherited trait in all generations without skipping
4) all of the above are correct

8. AUTOSOSOME RECESSIVE TYPE OF INHERITANCE IS CHARACTERIZED IN THAT

1) the ratio of healthy and sick family members is 1:1
2) the disease is not related to consanguinity
3) parents of the first identified patient are clinically healthy
4) all of the above is wrong

9. GENOMIC MUTATIONS ARE

1) Violation in the structure of the gene
2) Change in the number of chromosomes
3) Accumulation of intron repeats
4) Change in the structure of chromosomes

10. DELETION IS IT

1) Genomic mutation
2) gene mutation
3) Chromosomal mutation

11. THE REPLACEMENT OF INDIVIDUAL NUCLEOTIDES IN THE DNA CHAIN WITH OTHERS IS RELATED TO

1) Chromosomal mutations
2) Genomic mutations
3) Gene mutations

12. UNDER WHAT TYPE OF INHERITANCE IS PATIENTS SIGNIFICANTLY MUCH MORE BORN IN FAMILIES WITH CONNECTIVE MARRIAGES

1) X-linked recessive
2) autosomal recessive
3) X-linked dominant

13. BASIC LAW OF POPULATION GENETICS - LAW

1) mendel
2) Beadle Tatum
3) Hardy- Weinberg
4) Morgana
5) Wright

14. THE CAUSE OF PATAU SYNDROME IS

1) having three copies of chromosome 13
2) unbalanced robertsonian a translocation that results in two normal copies of chromosome 13 and an extra long arm of chromosome 13
3) mosaicism , which results in 3 copies of chromosome 13 in some cells and 2 copies in others
4) all of the above

15. BALANCED STRUCTURAL ANOMALIES ARE

1) gain or loss of genetic material
2) rearrangement of genetic material, but no overall gain or loss
3) break within a chromosome
4) a break within a chromosome, the gain or loss of genetic material

16. ROBERTSONIAN TRANSLOCATIONS ARE

1) movement of a segment from one chromosome to another without a noticeable increase or loss of DNA
2) movement of a segment from one chromosome to another, with a change in the total amount of DNA
3) a type of chromosomal rearrangement that is formed by the fusion of entire long arms of two acrocentric chromosomes (chromosomes with a centromere near the very end)
4) a type of chromosomal rearrangement that is formed by the fusion of the entire long arms of two chromosomes

17. CAN A PARENT WITH A BALANCED ROBERTSON TRANSLOCATION T(21Q;21Q) HAVE HEALTHY CHILDREN WITH RESPECT TO DOWN SYNDROME

1) yes, in 25% of cases
2) yes, in 50% of cases
3) yes, in 75% of cases
4) No

18. CYTOGENETIC METHODS FOR DIAGNOSTICS OF CHROMOSOMAL DISORDERS INCLUDED

1) karyotyping

2) chromosomal microarray analysis 3) fluorescent hybridization in situ (FISH) 4) all of the above	
19. ANEUPLOIDS IS 1) an increase in the entire set of chromosomes, a multiple of the haploid 2) an increase in one pair of chromosomes 3) exchange of regions between two pairs of homologous or non-homologous chromosomes 4) insertion of genetic material into a chromosome	
20. MOSAICISM IS 1) a condition in which a person has chromosomal abnormalities in some but not all cells 2) a condition in which a person has chromosomal abnormalities in all cells 3) a condition in which a person has an increase in the entire set of chromosomes that is a multiple of the haploid 4) exchange of regions between two pairs of homologous or non-homologous chromosomes	

test item number	response standard number
1.	2
2.	4
3.	2
4.	3
5.	4
6.	1
7.	4
8.	4
9.	1
10.	3
eleven.	3
12.	2
13.	3
14.	4
15.	2
16.	3
17.	4
18.	4
19.	2
20.	1

6. Criteria for evaluating learning outcomes

For the credit

Learning outcomes	**Evaluation criteria**	
	Not passed	**Passed**

Completeness of knowledge	The level of knowledge is below the minimum requirements. There were bad mistakes.	The level of knowledge in the volume corresponding to the training program. Minor mistakes may be made
Availability of skills	Basic skills are not demonstrated when solving standard tasks. There were bad mistakes.	Basic skills are demonstrated. Typical tasks have been solved, all tasks have been completed. Minor mistakes may be made.
Availability of skills (possession of experience)	Basic skills are not demonstrated when solving standard tasks. There were bad mistakes.	Basic skills in solving standard tasks are demonstrated. Minor mistakes may be made.
Motivation (personal attitude)	Educational activity and motivation are poorly expressed, there is no willingness to solve the tasks qualitatively	Educational activity and motivation are manifested, readiness to perform assigned tasks is demonstrated.
Characteristics of competence formation*	The competence is not fully formed. The available knowledge and skills are not enough to solve practical (professional) tasks. Repeated training is required	The competence developed meets the requirements. The available knowledge, skills and motivation are generally sufficient to solve practical (professional) tasks.
The level of competence formation*	Low	Medium/High

For testing:

Mark "5" (Excellent) – points (100-90%)
Mark"4" (Good) – points (89-80%)
Mark "3" (Satisfactory) – points (79-70%)

Less than 70% – Unsatisfactory – Mark "2"

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