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

FUNDAMENTALS OF MEDICAL GENETICS

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

Department: **BIOLOGY**

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 "Fundamentals of Medical genetics" is an integral appendix to the working program of the discipline " Fundamentals of Medical 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:

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 for measuring the level of knowledge and skills of a student	Bank of test tasks
2	Case-task	A problem task in which the student is offered to comprehend a real professionally-oriented situation necessary for solving	Tasks for solving cases

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

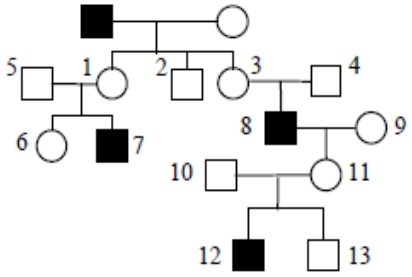
Code and formulation of competence*	Stage of competence formation	Controlled sections of the discipline	Assessment tools
EK-1 The student is able to carry out a critical analysis of problem situations based on a systematic approach, to develop a strategy of actions	Entry, Current, Mid-term	**Section 1.** Molecular genetics-the basis of medical genetics. Genetic passport. Epigenetics. Ethnogenomics.	Test control
		Section 2. Methods of studying human genetics. Monogenic, chromosomal and genomic diseases.	1.Test control 2. Case assignment
		Section 3. Congenital diseases and malformations.	Test control
		Section 4. Medical and genetic counseling as a type of specialized medical care for the population.	Test control

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

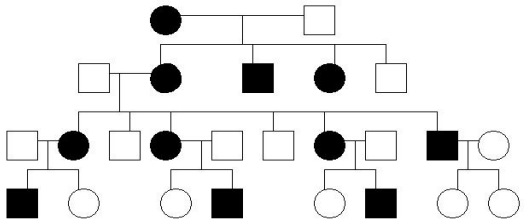
The current control is carried out by the discipline teacher when conducting classes in the form of: testing, case assignments.

4.1. Case assessment tools of competence EC-1

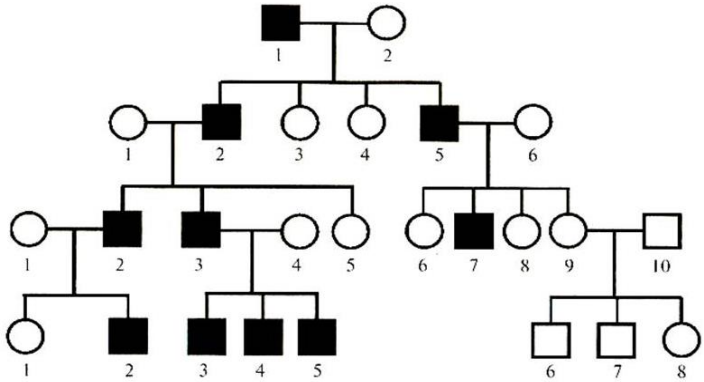
1. Determine the nature of the inheritance of the trait, arrange the genotypes of all members of the pedigree.



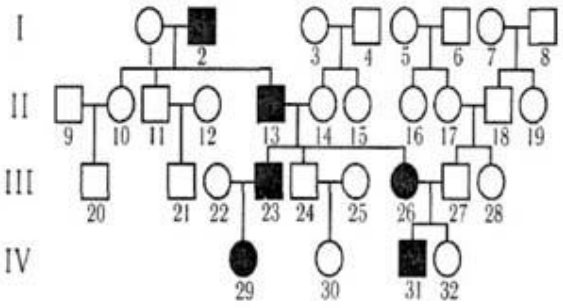
2. Determine the nature of the inheritance of the trait, arrange the genotypes of all members of the pedigree.



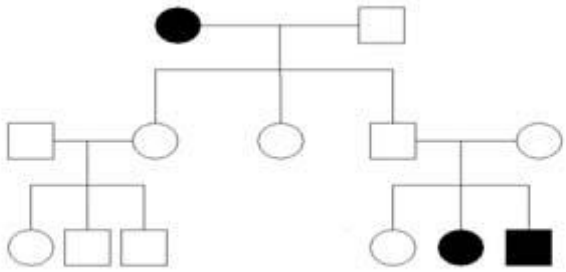
3. Determine the nature of the inheritance of the trait, arrange the genotypes of all members of the pedigree.



4. Determine the nature of the inheritance of the trait and arrange the genotypes of all members of the pedigree.



5. Determine the nature of the inheritance of the trait, the genotypes of children in the first and second generation.



6. It is known that in the population of phenylketonuria occurs with a frequency of 1:10000. Phenylketonuria is inherited by the autosomal recessive type (aa). Find the frequency of occurrence of heterozygotes (Aa).

7. In Europeans, Rh-negativity occurs with a frequency of 16% and is inherited autosomal recessive. Find the frequency of occurrence of heterozygotes.

8. Albinism is inherited as an autosomal recessive trait. The disease occurs with a frequency of 1:20000. Find the frequency of occurrence of heterozygotes.

9. Hemophilia is inherited as a recessive sex-linked X-chromosome trait. In newborn boys, this disease occurs with a frequency of 1:2500. Determine the frequency of occurrence of heterozygotes among newborns.

10. 2 cases of phenylketonuria were registered in the city with 2000 inhabitants. Determine the number of heterozygotes in the population.

11. Gout occurs in 2% of people and is caused by an autosomal dominant gene. In women, gout does not manifest itself, in men, the penetration rate is 20%. Determine the genotypic structure of the population based on the analyzed trait.

5. The content of the assessment tools of mid-term assessment

Mid-term assessment is carried out in the form of a credit.

The content of the assessment tool (questions, topics of abstracts, round tables, etc.)

If the bank of assessment tools for conducting current control and mid-term assessment of students in this discipline is presented on the Educational Portal of the PRMU, specify a link to this electronic resource.

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

5.1.1. Questions for the credit in the discipline "Medical genetics".

Question	Competence code (according to the WPD)
1. Arrange the main stages of DNA research in the correct order 1) Conducting a DNA study in accordance with the chosen method 2) Collection and interpretation of data 3) Taking off material for molecular genetic research 4) Isolation of DNA or RNA from a sample	
2. Put the PCR steps in the correct order 1) Annealing 2) Elongation 3) Denaturation	
3. Are the blunt or sticky end patterns of cleavage by restriction endonucleases are shown in the figure? 1) Sticky-ended pattern of cleavage by restriction endonucleases **2)** Blunt-ended pattern of cleavage by restriction endonucleases	
4. **Are the blunt or sticky end patterns of cleavage by restriction endonucleases are shown in the figure?** 1) Sticky-ended pattern of cleavage by restriction endonucleases **2)** Blunt-ended pattern of cleavage by restriction endonucleases	

5. Name the main components of DNA 1) Deoxyribose 2) Nitrogen-containing nucleobases (cytosine, guanine, adenine, uracil) 3) Phosphate group 4) Ribose 5) Nitrogen-containing nucleobases (cytosine, guanine, adenine, thymine)	
6. Name the main components of RNA 1) Deoxyribose 2) Nitrogen-containing nucleobases (cytosine, guanine, adenine, uracil) 3) Phosphate group 4) Ribose 5) Nitrogen-containing **nucleobases (cytosine, guanine, adenine, thymine)**	
7. Select the required components for PCR 1) A DNA sample 2) Two DNA primers 3) DNA polymerase (Taq-polymerase) 4) A buffer solution **5)** Deoxynucleoside triphosphates	
8. What are restriction endonucleases? 1) These are bacterial enzymes that cut double-stranded DNA into specific sequences. 2) These are fragments of variable-length DNA or RNA that can be radioactively or fluorescently labeled. **3)** These are sections of DNA that are formed as a result of cutting by endonucleases.	
9. What are the main conditions for taking off material for molecular genetic research? 1) Samples for DNA research are taken only with a disposable instrument. 2) Samples for DNA research are taken only in disposable test tubes. 3) Samples for DNA research are taken only with a disposable instrument in disposable test tubes. 4) Samples for DNA research are taken only in sterile conditions.	
10. What bond joined the nucleotides to one another in a chain? 1) Peptide bond 2) Hydrogen bonding 3) Glycosidic bond 4) Phospho-diester bond 5) Hydrogen sulfide bond	
11. What DNA methods are modifications of nucleic acid hybridization? 1) Southern blotting 2) Polymerase chain reaction 3) DNA chip assay 4) Fluorescence in situ hybridization 5) DNA sequencing	
12. What enzyme is needed for PCR? 1) Restriction endonuclease 2) Taq-polymerase 3) DNA topoisomerase 4) RNA polymerase 5) Revertase	
13. What enzyme is needed for RFLP? 1) Restriction endonuclease 2) Taq-polymerase 3) DNA topoisomerase	

4) RNA polymerase 5) Revertase	
14. What enzyme is needed for the Southern-blotting method? 1) RNA polymerase 2) Taq-polymerase 3) DNA topoisomerase 4) Restriction endonuclease 5) Revertase	
15. What fragments of DNA are called microsatellites? 1) These are short (1-4 base pairs) highly repetitive DNA fragments. **2)** These are long (5-64 base pairs)highly repetitive DNA fragments that are concentrated near the centromeres and telomeres.	
16. What fragments of DNA are called minisatellites? 1) These are long (5-64 base pairs)highly repetitive DNA fragments that are concentrated near the centromeres and telomeres. 2) These are short (1-4 base pairs) highly repetitive DNA fragments.	
17. What is a DNA probe? 1) It is a fragment of DNA or RNA of variable length which can be radioactively or fluorescently labeled. 2) It is a bacterial enzyme that cut double-stranded DNA into specific sequences. 3) These are sections of DNA that are formed as a result of cutting by endonucleases.	
18. What is the charge of nucleic acid molecules in an aqueous solution? 1) Neutral 2) Positive 3) Negative	
19. What is the essence of the electrophoresis method? 1) The motion of charged molecules under the action of an electric field. 2) The movement of uncharged molecules. 3) The movement of charged molecules under the action of light.	
20. What method is based on a conformational difference of single-stranded nucleotide sequences? 1) Single strand conformation polymorphism 2) DNA sequencing 3) Polymerase chain reaction 4) Nucleic acid hybridization 5) DNA profiling	
21. A method used more for mapping and cloning genes involved in carcinogenesis 1) SKY - spectral karyotyping; 2) CGH - comparative genomic hybridization; 3) M-FISH -(multicolor or multiplex FISH); 4) LSI - locus specific identificator;	
22. Aneuploidy 1) Genomic mutation; 2) Chromosomal mutations; 3) A change in the karyotype in which the number of chromosomes in the cells is not a multiple of the haploid set; 4) A point mutation that does not affect the phenotype**;**	
23. Biochemical method 1) Study of the peripheral blood cell culture; 2) Studies of patterns on the palms, soles and fingers; 3) Drawing up a pedigree for inheritance analysis;	

4) Detection of defects in enzymes, structural proteins; 5) Study of inheritance in pairs of identical and fraternal twins	
24. C-staining is used: 1) For the study of Y-chromosomes; 2) To detect small aberations and marker chromosomes; 3) To identify details of homologous G-or Q-negative sites of sister chromatids or homologous chromosomes; 4) For the analysis of centromeric regions of chromosomes; 5) For the analysis of telomeric regions of chromosomes	
25. Chemical differences detected by differential staining methods 1) Q-segments correspond to sections rich in A-T-pairs; 2) The presence of dividing cells; 3) Identification of a number of translocations (bcr/abl, MLL, PML/RARA, TEL/AML1); 4) R-segments correspond to cross-sections rich in G-C-pairs	
26. Colcemide (or colchicine) is used 1) To stimulate cell growth in culture; 2) To block dividing cells at the metaphase stage; 3) For the analysis of a state of proteins; 4) As a fluorescent marker	
27. Combination variation 1) The appearance of new signs with a random combination of gametes; 2) Changes in the number of chromosomes; 3) Changes in the structure of the gene; 4) Changes in the structure of chromosomes during crossing-over;	
28. Concordance 1) An emotionally stable person, calm and balanced; 2) Percentage of difference in the studied trait; 3) Percentage of similarity in the studied feature;	
29. Cytogenetic method 1) The object of the study is the culture of peripheral blood cells; 2) Study of inheritance in pairs of identical and fraternal twins; 3) Drawing up a pedigree for inheritance analysis; 4) Detection of defects in enzymes, structural proteins; 5) All of the above;	
30. Down Syndrome 1) Karyotype 46,XXY; 2) Karyotype 47,XX/XY, +21; 3) Karyotype 46,XXX; 4) Karyotype 48,XXY;	
31. Edwards Syndrome 1) Karyotype 47,XX/XY, +18; 2) Karyotype 47,XX/XY,+21; 3) Karyotype 46,XXX; 4) Karyotype 48,XXY;	
32. G-staining is used 1) For the study of Y-chromosomes; 2) To detect small aberations and marker chromosomes; 3) To identify details of homologous G-or Q-negative sites of sister chromatids or homologous chromosomes; 4) For the analysis of centromeric regions of chromosomes; 5) For analysis of telomeric regions of chromosomes;	
33. Hereditary diseases 1) Diseases transmitted only from parents to descendants;	

2) Pathological conditions caused by a change in the genetic material; 3) Diseases of children from healthy parents; 4) All of the above;	
34. If a the sign appears in one of the twins, then the pair is called 1) Monozygotic; 2) Concordant; 3) Disconcordant; 4) Dizygotic;	
35. Immunological methods are used 1) For the quantitative determination of sugar in the urine; 2) A microbiological test used to detect certain hereditary metabolic disorders; 3) When the chromosomes are fluorescently colored; 4) When determining the number and structure of chromosomes; 5) In case of suspected antigenic incompatibility of the mother and fetus;	
36. Insertion 1) The loss of nucleotide; 2) Nucleotide insertion; 3) Replacement of nucleotides; 4) Rearrangement of nucleotides;	
37. Inversion 1) A shift in the reading frame; 2) Replacement of nucleotides; 3) Overturning a DNA fragment; 4) All of the above;	
38. Klinefelter syndrome 1) Karyotype 47,XX/XY,+18; 2) Karyotype 45,XO; 3) Karyotype 47,XXY; 4) Karyotype 47,XX/XY,+13;	
39. Missense mutation 1) Replacement of the nucleotide in the codon, resulting in the encoding of the stop codon; 2) Replacement of a nucleotide in a codon, resulting in the encoding of another amino acid; 3) Replacement of the nucleotide in the codon, leading to a shift in the reading frame; 4) All of the above;	
40. Mutations 1) Changes in the nucleotide composition; 2) Changes in the number of chromosomes; 3) Changes in the structure of the gene; 4) All of the above;	
41. Necessary conditions for cytogenetic diagnosis 1) Use of fluorescent markers; 2) Use of colcemide (or colchicine); 3) Hypotonic shock; 4) The presence of a proband; 5) The presence of dividing cells;	
42. Nonsense-mutation 1) Genomic mutation; 2) Chromosomal mutations; 3) Point mutation leading to the formation of a stop codon; 4) A point mutation that does not affect the phenotype;	

43. Patau syndrome 1) Karyotype 47,XX/XY,+18; 2) Karyotype 47,XX/XY,+21; 3) Karyotype 46,XXX; 4) Karyotype 47,XX/XY,+13;	
44. Amniocentesis is 1) obtaining of a small amount of placental tissue; 2) obtaining of amniotic fluid with fetal cells; 3) fetal blood sampling; 4) obtaining of fragments of fetal genetic material contained in the mother's blood;	
45. Cordocentesis is 1) obtaining of a small amount of placental tissue; 2) obtaining of amniotic fluid with fetal cells; 3) fetal blood sampling; 4) obtaining of fragments of fetal genetic material contained in the mother's blood;	
46. In prenatal diagnostics, the following methods are distinguished: 1) extra-corporal fertilization; 2) invasive; 3) non-invasive; 4) screening tests;	
47. The cell-free DNA (cfDNA) method is based on: 1) obtaining of DNA from fetal cells; 2) the analysis of fragments of fetal genetic material contained in the mother's blood; 3) obtaining of DNA from the mother's cells; 4) all of the above;	
48. The contraindications for invasive prenatal diagnostics are 1) detachment of the placenta; 2) the age of the mother is over 35 years old; 3) a risk of termination of pregnancy; 4) a mother's a carrier of the X-linked recessive disease gene; 5) abnormal development of the uterus;	
49. The indications for invasive prenatal diagnostics are 1) the age of the mother is over 35 years old; 2) the presence of structural rearrangements of chromosomes in one of the parents; 3) a risk of termination of pregnancy; 4) abnormal development of the uterus; 5) a mother's a carrier of the X-linked recessive disease gene; 6) all of the above;	
50. The invasive methods include: 1) amniocentesis; 2) ultrasound and serological tests; 3) cordocentesis; 4) examination of a fetus without surgery; 5) biopsy of skin of a fetus;	

Test task №	response standard №	Test task №.	response standard №	Test task №.	response standard №
1	3412	21	2	41	2,3,5
2	231	22	1,3	42	3

3	1	23	4	43	4
4	2	24	4	44	2
5	1,3,3	25	1,4	45	3
6	2,3.4	26	2	46	1,2
7	1,2,3,4,5	27	1	47	2
8	1	28	3	48	1,3,5
9	3	29	1	49	1,2,5
10	4	30	2	50	1,3,5
11	1,3,4	31	1		
12	2	32	2		
13	1	33	2		
14	4	34	3		
15	1	35	5		
16	1	36	2		
17	1	37	3		
18	3	38	2		
19	1	39	2		
20	1	40	4		

Coursework as an element of an academic discipline should contribute to the formation of competencies provided for in the competence matrix for this discipline and specified in the WPD**.**

6. Criteria for evaluating learning outcomes

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"

A complete set of evaluation tools for the discipline "Medical genetics" is presented on the portal of the Volga Research Medical University – link *https://sdo.pimunn.net/course/view.php?id=2015*

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Date "_____" _____ 202__