

Документ подписан простой электронной подписью

Информация о владельце:

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Должность: и.о. декана медико-биологического факультета

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MINISTRY OF SCIENCE AND HIGHER EDUCATION

RUSSIAN FEDERATION

Federal State Autonomous Educational Institution of Higher Education

NORTH-CAUCASUS FEDERAL UNIVERSITY

APPROVED

Assoc. Prof. Kubanov S.I.

PhD in Medical Sciences

Dean

Faculty of Medicine and Biology

ASSESSMENT TOOLS FUND FOR THE DISCIPLINE

Medical Genetics

- Specialty: 31.05.01 General Medicine
- Mode of training: Full-time
- Year of commencement of studies: 2026
- Delivered in Semester 9

Introduction

1. Intended for carrying out current control of academic performance progress and intermediate attestation in the discipline of Medical Genetics for students of the specialty (majoring in) 31.05.01 General Medicine.
2. The ATF is an appendix to the program of the discipline (module) Medical Genetics
3. Developer(s): Kukharuk Maksim Yurievich Docent of the Department of Genetics and Breeding
Baculina Elena Gennadyevna Docent of the Department of Genetics and Breeding
4. Expert examination of the ATF has been completed.

Expert group:

Chairperson: Pahomova Tatyana Aleksandrovna Docent of the Department of Anatomy and Histology

Committee members: Krivoruchko Aleksandr Yurievich, Head of of the Department of Genetics and Breeding

Zuev Roman Vladimirovich Docent of the Department of Genetics and Breeding

Employer organization representative: Vilensky I.L., Chief Physician of Independent CDL, LLC

Expert conclusion – the ATF allows evaluating the level of competency levels and is recommended to be employed in the educational process.

5. The validity period for the ATF shall be determined by the period of implementation of the educational program.

1. Description of Evaluation Criteria at Various Stages of Their Formation, Description of Evaluation Scales

Levels of formation of competence(s), indicator(s)	Descriptors			
	Minimum level not achieved (Unsatisfactory) 2 points	Minimum level (Satisfactory) 3 points	Intermediate level (Good) 4 points	High level (Excellent) 5 points
Competency PC-2. Ready to collect and analyze patient complaints, history data, examination results, laboratory, instrumental, pathological-anatomical, and other studies to recognize a condition or establish the fact of the presence or absence of a disease				
Learning outcomes for the discipline (module): Development of clinical thinking. Indicator: ID-1 PC-2 Knows how to carry out the collection of complaints, anamnesis of life, and disease of the patient and analyze the information obtained	Does not know how to carry out the collection of details concerning the patient's complaints, anamnesis of life, and disease	Knows how to carry out the collection of details concerning the patient's complaints, anamnesis of life, and disease	Knows how to carry out the collection of details concerning the patient's complaints, anamnesis of life, and disease	Knows how to carry out the collection of details concerning complaints, anamnesis of life, and disease, and how to analyze the information obtained
Learning outcomes for the discipline (module): Development of clinical thinking. Indicator: ID-2 PC-2 Conducts a complete physical examination of the patient (inspection, palpation, percussion, auscultation) and interprets its results	Does not carry out a complete physical examination of the patient	Carries out a complete physical examination of the patient	Carries out a complete physical examination of the patient (inspection, palpation, percussion, auscultation)	Carries out a complete physical examination of the patient (inspection, palpation, percussion, auscultation) and interprets its results

Learning outcomes for the discipline (module): Development of clinical thinking. Indicator: ID-3 PC-2 Defines a preliminary diagnosis and compiles a plan for laboratory and instrumental examinations of the patient	Does not define a preliminary diagnosis	Defines a preliminary diagnosis	Defines a preliminary diagnosis and compiles a plan for laboratory examinations	Defines a preliminary diagnosis and compiles a plan for laboratory and instrumental examinations
Competency PC-3. Capable of determining a patient's main pathological states, symptoms, disease syndromes, and nosological forms in accordance with the International Statistical Classification of Diseases and Related Health Problems, 10th Revision (ICD-10).				
Learning outcomes for the discipline (module): Acquisition of special medical skills and their application in practical healthcare. Indicator: ID-1 PC-3 Is capable of building an algorithm and selecting criteria for determining the main pathological states, syndromes, and diseases in patients of various profiles in accordance with the International Statistical Classification of Diseases and Related Health Problems, 10th Revision (ICD-10)	Not capable of building an algorithm and selecting criteria for identifying various profiles in the patient	Capable of building an algorithm and selecting criteria for identifying various profiles in the patient	Capable of building an algorithm and selecting criteria for identifying the patient's various profiles of main pathological conditions, syndromes, diseases	Capable of building an algorithm and selecting criteria for identifying the patient's various profiles of main pathological conditions, syndromes, diseases following the International Statistical Classification of Diseases and Related Health Problems, 10th Revision (ICD-10)

Learning outcomes for the discipline (module): Acquisition of special medical skills and their application in practical healthcare. Indicator: ID-2 PC-3 Identifies main pathological symptoms and syndromes, formulates a clinical diagnosis in accordance with ICD-10	Does not identify main pathological symptoms	Identifies main pathological symptoms	Identifies main pathological symptoms and syndromes, defines a clinical diagnosis	Identifies main pathological symptoms and syndromes, defines a clinical diagnosis subject to ICD-10
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The evaluation of the competency formation level in the discipline is carried out on the basis of the Regulations on conducting current control of academic progress and intermediate attestation of students in higher education educational programs – Bachelor's programs, Specialist's programs, Master's programs – at the Federal State Autonomous Educational Institution of Higher Education NORTH-CAUCASUS FEDERAL UNIVERSITY in its current edition.

ASSESSMENT TOOLS FOR CHECKING THE LEVEL OF COMPETENCY DEVELOPMENT

Task Number	Correct Answer	Content of the Question	Competency
1	c	The "bottleneck effect" is: a) an increase in the number of mutants; b) a change in gene position on the chromosome; c) random death of carriers of a particular genotype with a significant reduction in population size; d) gene exchange between different populations.	PC-2
2	b	In prokaryotes, genes are grouped into: a) Operator; b) Operon; c) Regulator; d) Replicon	PC-2
3	e	A newborn presents with hypotonia, a single transverse palmar crease, epicanthic folds, and a congenital heart defect. A karyotype reveals an extra chromosome 21. Which of the following is the correct karyotype notation? a) 45, X b) 47, XXY c) 47, XX+18	PC-2

		d) 47, XY+13 e) 47, XX+21	
4	c	A couple has a child affected with cystic fibrosis (autosomal recessive). Both parents are phenotypically healthy. What is the probability that their next child will be a carrier (heterozygote) of the cystic fibrosis gene? a) 0% b) 25% c) 50% d) 75% e) 100%	PC-2
5	d	The following traits are not inherited by cumulative polymerism: a) milk production, egg production, weight; b) parameters of physical strength and mental abilities in humans; c) ear length, sugar content; d) eye color.	PC-3
6	c, e, g	Identify several diseases that are in one way or another associated with sex chromosomes: a) Down syndrome; b) Phenylketonuria; c) Klinefelter syndrome; d) Hemophilia; e) Patau syndrome; f) Shereshevsky-Turner syndrome; g) Color blindness; h) Edwards syndrome.	PC-3
7	c	A healthy woman whose father was affected with hemophilia A (X-linked recessive) is planning a pregnancy. She is a carrier of the mutation. What is the probability that her son will be affected with hemophilia? a) 0% b) 25% c) 50% d) 75% e) 100%	PC-3
8	c	A pregnant woman at 16 weeks of gestation undergoes amniocentesis due to advanced maternal age (39 years). The fetal karyotype shows 47, XY+13. Which syndrome does the fetus have? a) Down syndrome b) Edwards syndrome c) Patau syndrome d) Klinefelter syndrome e) Turner syndrome	PC-3
9	c	A patient is suspected of having Huntington's disease, which is caused by an expanded CAG trinucleotide repeat in the HTT gene. Which molecular technique is most commonly used to confirm the diagnosis? a) Southern blot	PC-2

		b) Karyotyping c) PCR fragment analysis (repeat sizing) d) Western blot e) Fluorescence in situ hybridization (FISH)	
10	1-C, 2-A, 3-B, 4-D, 5-E	Match each syndrome with its correct karyotype 1. Down syndrome 2. Turner syndrome 3. Klinefelter syndrome 4. Edwards syndrome 5. Patau syndrome A. 45, X B. 47, XXY C. 47, XX+21 (or 47, XY+21) D. 47, XX+18 (or 47, XY+18) E. 47, XX+13 (or 47, XY+13)	PC-2
11	1-B, 2-E, 3-C, 4-A, 5-D	Match each gene with its associated drug or clinical response 1. CYP2C9 2. VKORC1 3. TPMT 4. CYP2D6 5. CYP2C19 A. Codeine metabolism (ultra-rapid metabolizers → toxicity) B. Warfarin dosing (sensitivity / resistance) C. Thiopurine toxicity (azathioprine, 6-mercaptopurine) D. Clopidogrel activation (poor metabolizers → reduced efficacy) E. Warfarin dosing (combined with VKORC1)	PC-2
12	1-D, 2-A, 3-B, 4-C, 5-E	Match each molecular genetic technique with its primary clinical application 1. PCR (fragment analysis / repeat sizing) 2. Karyotyping 3. Sanger sequencing 4. FISH (fluorescence in situ hybridization) 5. Next-generation sequencing (NGS) A. Detection of chromosomal aneuploidies in prenatal samples (amniocentesis, CVS) B. Detection of point mutations and small deletions (e.g., CFTR, BRCA1/2) C. Detection of specific DNA or RNA sequences in tissue sections (e.g., HER2, ALK) D. Diagnosis of trinucleotide repeat disorders (e.g., Huntington's, fragile X) E. Comprehensive testing for germline or somatic mutations in multiple genes (e.g., cancer panels, exome sequencing)	PC-2
13	1-C, 2-A, 3-B, 4-D, 5-E	Match each genetic disorder with its primary molecular mechanism or type of mutation 1. Cystic fibrosis 2. Huntington's disease 3. Sickle cell anemia	PC-3

		4. Fragile X syndrome 5. Duchenne muscular dystrophy A. Trinucleotide repeat expansion (CAG) B. Point mutation in the β -globin gene (hemoglobin S) C. Deletion of one or more nucleotides (e.g., F508del in CFTR) D. Trinucleotide repeat expansion (CGG) in the 5' UTR of FMR1 E. Large deletions or frameshift mutations in the dystrophin gene (DMD)	
14	1-C, 2-A, 3-D, 4-E, 5-B	Match each inheritance pattern with its clinical feature 1. Autosomal dominant 2. Autosomal recessive 3. X-linked recessive 4. X-linked dominant 5. Y-linked (holandric) A. Affected individuals are usually born to unaffected carrier parents; both sexes affected equally B. Trait appears only in males; father transmits to all sons C. Trait appears in every generation; affected males and females are equally represented; male-to-male transmission occurs D. Affected males are more frequent; no male-to-male transmission; affected males inherit the mutation from their mother E. Both sexes affected; affected males transmit the trait to all daughters but to no sons	PC-3
15	L1) R3; L2) R1; L3) R2;	Match the syndrome and its Phenotypic manifestation: L1) Phenylketonuria; L2) Marfan syndrome; L3) Tyrosinemia. R1) Tall stature, long "spider" fingers; R2) Insufficient skin pigmentation, corneal opacity; R3) Impaired muscle tone and reflexes, seizures.	PC-3
16	L1 R2; L2 R3; L3 R1	Match the types of sex determination: L1) Progametic; L2) Syngametic; L3) Epigametic. R1) formed under the influence of the environment; R2) before fertilization, by the structure of male and female gametes; R3) genetic sex determination at fertilization, depending on the combination of sex chromosomes or the ratio of sex chromosomes to autosomes.	PC-3
17	1-C, 2-B, 3-A, 4-D.	Arrange in order the stages of sex formation in humans: A) phenotypic sex; B) gonadal sex determination; C) genetic sex determination; D) behavioral sex.	PC-2
18	1-D, 2-C,	Arrange the following steps of genetic counseling in the correct chronological order	PC-2

	3-E, 4-B, 5-A	A. Provide follow-up support and psychological assistance B. Discuss reproductive options (prenatal diagnosis, PGD, adoption, etc.) C. Establish the diagnosis and determine the mode of inheritance D. Take a family history and construct a pedigree E. Estimate the recurrence risk for the family	
19	1-C, 2-E, 3-B, 4-A, 5-D	Arrange the following steps of an amniocentesis procedure in the correct chronological order A. Culture the fetal cells for karyotyping or molecular analysis B. Collect amniotic fluid (15–20 ml) by transabdominal aspiration C. Perform ultrasound to locate the fetus and identify a pocket of amniotic fluid D. Analyze the results (karyotype, FISH, or DNA analysis) and counsel the patient E. Insert a sterile needle through the maternal abdomen into the amniotic cavity	PC-2
20	1-D, 2-B, 3-E, 4-A, 5-C	Arrange the following steps in the clinical diagnostic workup for a patient suspected of having a genetic disorder in the correct chronological order A. Order and interpret confirmatory molecular genetic tests (e.g., PCR, sequencing, NGS panel) B. Perform a detailed physical examination (dysmorphology, neurological, etc.) C. Provide genetic counseling to the patient and family D. Take a thorough family history and construct a three-generation pedigree E. Evaluate laboratory and imaging findings (biochemical, radiological)	PC-2
21	1-B, 2-E, 3-D, 4-A, 5-C	Arrange the following steps for analyzing a pedigree with a suspected mitochondrial disorder in the correct chronological order A. Trace maternal lineage through the pedigree (all children of affected mothers inherit the mutation) B. Collect family history and construct a pedigree focusing on maternal transmission C. Perform molecular testing for mtDNA mutations (e.g., point mutations, deletions) D. Evaluate clinical symptoms (e.g., neurological, muscular, visual, hearing impairment) E. Consider mitochondrial inheritance when affected individuals show variable expressivity and maternal transmission	PC-3
22	1-D, 2-B, 3-C, 4-A, 5-E	Arrange the following events of apoptosis in the correct chronological order A. Cell breaks into apoptotic bodies B. Activation of executioner caspases (caspase-3, -6, -7) C. DNA fragmentation and nuclear condensation D. Release of cytochrome c from mitochondria	PC-3

		E. Phagocytosis of apoptotic bodies by macrophages or adjacent cells	
23	1-B, 2-A, 3-D, 4-C	Arrange the following steps of a single PCR cycle in the correct chronological order A. Primer annealing (primers bind to complementary template sequences) B. Denaturation of double-stranded DNA into single strands C. Final extension completes the synthesis of all products (after the last cycle) D. Elongation (DNA polymerase extends primers, synthesizing new DNA strands)	PC-3
24	arachnodactyly	A characteristic feature of Marfan syndrome is ###	PC-3
25	Etiological	A treatment method that changes the cause of a hereditary disease and radically cures it is: ###	PC-2
26	25% 0,25	A colorblind girl has a mother and a brother with normal vision, as well as a colorblind brother. What is the probability that her parents will give birth to a girl with normal vision?	PC-2
27	Trisomy	The detection of two Barr bodies (sex chromatin clumps) in somatic cell nuclei of a female organism indicates a syndrome of ###	PC-3
28	Discordance	The degree of difference between twins is called ###	PC-3
29	Cytogenetic	The ### research method in genetics allows for the study of the karyotype of cells and the detection of genomic and chromosomal mutations.	PC-3
30	Parthenogenesis	A type of sexual reproduction in which female reproductive cells develop without fertilization? ###	PC-3

2. Description of the Evaluation Scale

Within the framework of the rating system, students' academic progress in each discipline is evaluated during current control and intermediate attestation. The rating system for assessing students' knowledge is based on the use of a combination of control activities to check the covered material (checkpoints), optimally located throughout the entire time interval of studying the discipline. The principles of the rating system for assessing students' knowledge are based on the requirements described in the Regulations on the organization of the educational process based on the rating system for assessing students' knowledge at NCFU.

3. Criteria for Evaluating Competencies

The grade **Excellent** shall be awarded to a student if the theoretical content of the course has been mastered completely, without gaps; the material is presented exhaustively, consistently, clearly, and logically; the student freely copes with tasks, questions, and other types of knowledge application; uses additional material in the answer; all tasks provided for by the program are completed, and the quality of their execution is evaluated by a number of points close to the maximum; analyzes the obtained results; demonstrates independence when performing tasks.

The grade **Good** is to be awarded to a student if the theoretical content of the course is mastered completely, the necessary practical competencies are mainly formed, all educational tasks provided for by the training program are completed, and the quality of their execution is sufficiently high. The student firmly knows the material, presents it competently and strictly on the merits of the matter, without allowing significant inaccuracies in answering the question.

The grade **Satisfactory** is awarded to a student in case the theoretical content of the course has been mastered in part only, but the gaps are not of a significant nature, most of the tasks provided for by the program are completed, but contain errors, the student allows inaccuracies and insufficiently correct wording when answering the question, and violations of logical sequence are observed in the presentation of the program material.

The grade **Unsatisfactory** is awarded to a student if they do not know a significant part of the program material, allow significant errors, perform practical works with great difficulty and uncertainty, the necessary practical competencies are not formed, most of the educational tasks provided for by the training program are not completed, and the quality of their execution is evaluated by a number of points close to the minimum.