Al-Farabi Kazakh National University Faculty of Biology and Biotechnology Department of Molecular Biology and Genetics

FINAL EXAMINATION PROGRAM BY SUBJECT

104830-Molecular and forensic-medical expertise Educational program "7M05105 - Genetics" 2024-2025 academic years spring semester

Course 1
Semester 2
Credits number 5

The final exam program was developed by associate professor of the Department of Molecular Biology and Genetics, Ph.D. Taipakova S.M. according to the main curriculum of the educational program "7M05105 - Genetics".

It was considered and presented at the meeting of the Department of Molecular Biology and Genetics.

"13 " March 2025 protocol No. 16.

Head of the department, Ph.D.,

Associate Professor Zhunusbaeva J.K.

BRIEF DESCRIPTION OF THE COURSE

Course Type: The course "101584 - Molecular and Forensic Examination" is designed for students specializing in Genetics. The course discusses the application of modern molecular-genetic methods in forensic examination, including criminology and the detection of genetically modified products, along with their advantages and specific features.

Final Exam Format: Written/offline (Traditional - Answering Questions)

The final exam questions cover the tasks studied throughout the semester in the "101584 - Molecular and Forensic Examination" course, including cognitive, functional, and systemic competency questions.

TOPICAL PLAN OF THE COURSE

- •Introduction to Forensic Medicine: History, Subject, and Tasks. History and development of forensic medicine. The subject and tasks of forensic medicine. The relationship of forensic medicine with other scientific disciplines
 - •Biological Sources of DNA: The Target Materials for Forensic DNA Typing
- •DNA sample sources. Biological evidence at crime scenes. Evidence collection and preservation. Collection of biological stains. Reference sample collection for DNA comparison and storage. DNA sample storage and preservation. Sample characterization. Main destructive influences on DNA.Withdrawal of samples out of storage.
 - •DNA Extraction and Quantitation of Forensic Samples
- •DNA extraction. General principles of DNA extraction. Quantification of DNA
 - •DNA extraction from challenging samples:
- •Briefly discuss the challenges of isolating DNA from degraded, old, or poorquality samples
- •Evaluate the efficacy of different extraction methods and analyze factors affecting DNA yield and quality
- •Polymerase Chain Reaction (PCR) as the Primary Method for DNA Analysis in Forensic Medicine
- •Principles of PCR. Classical PCR vs. new approaches: qPCR and multiplex PCR.
- •The evolution of PCR-based profiling in forensic genetics. DNA replication the basis of the PCR. The components of PCR. The PCR process. PCR inhibition. Sensitivity and contamination. The PCR laboratory
 - •The analysis of short tandem repeats
- •Analysis of tandem repeats and DNA polymorphisms. Structure of STR loci. The development of STR multiplexes. Detection of STR polymorphisms. Interpretation of STR profiles.

- •Assessment of STR profiles. Stutter peaks. Split peaks (+/-A). Pull-up Template DNA. Overloaded profiles. Low copy number DNA. Peak balance. Mixtures. Degraded DNA
- •Fundamentals of DNA Separation and Detection: DNA electrophoresis. DNA separation mechanisms. DNA fragment sizing and resolution. Methods for detecting DNA molecules
 - •DNA Fingerprinting and Genetic Identification of Individuals
 - •Statistical Interpretation: Evaluating the Strength of Forensic DNA Evidence
- •Population genetics. Deviation from the Hardy—Weinberg equilibrium. Statistical tests to determine deviation from the Hardy—Weinberg equilibrium. Estimating the frequencies of STR profiles. Corrections to allele frequency databases. Which population frequency database should be used?
 - •DNA typing by Restriction Fragment Length Polymorphism
- •Introduction to RFLP. Discovery of RFLP. Principle of RFLP. Restriction Endonuclease. Genetic variations. Detection of RFLP
 - •The analysis of single nucleotide polymorphisms.
- •SNPs occurrence and structure. Detection of SNPs. SNP detection for forensic applications. Forensic applications of SNPs. SNPs compared to STR loci
 - •DNA Databases: Databases of DNA profiles, Database vs. Databank
 - •The X Chromosome in Forensic Science: Past, Present and Future
- •Forensic utilization of the X chromosome. Chromosome X short tandem repeats. Power of ChrX markers in trace analysis. Power of ChrX markers in kinship testing. Chromosome X marker mapping and haplotype analysis. Chromosome X—chromosome Y homologue markers. Chromosome X STR allele and haplotype distribution in different populations. Ethical considerations in ChrX marker testing
- •Mitochondrial DNA as a Molecular-Genetic Object for Identifying Individuals
- •Molecular-genetic analysis of mitochondrial DNA. Application of mitochondrial DNA in forensic identification. Sequencing mitochondrial DNA and its application in forensic practice
- •Modern DNA Sequencing Methods in Forensic Practice. "Human Genome" project and next-generation sequencing (NGS) methods. Advantages and perspectives of NGS in forensic expertise. Practical use of NGS for DNA analysis. Comparison of traditional sequencing methods with NGS
- •The Y chromosome. Forensic applications of Y chromosome polymorphisms
- •Introduction to the Y Chromosome in Forensics. Structure and Function of the Y Chromosome. Y-STR Typing in Forensic Science. Forensic. Applications of Y Chromosome Polymorphisms. Y-SNP Analysis and Its Forensic Significance. Challenges and Limitations of Y Chromosome Analysis. Legal and Ethical Considerations in Y Chromosome Analysis
- •The Role of Molecular-Genetic Expertise in Investigating Sexual Crimes Against Minors

- •Application of molecular-genetic expertise in criminal investigations. Ethical and legal aspects of molecular-genetic methods. Case studies on the role of molecular-genetic expertise in sexual crime investigations
 - •DNA Phenotyping and Paternity Testing Methods
- •Principles and technologies of DNA phenotyping. Use of molecular-genetic methods for paternity testing. Application of DNA phenotyping methods for identifying physical traits
 - •Forensic Challenges: Degraded DNA, Mixtures, and LCN
- •Use of reduced-size PCR products (miniSTRs). Value of highly polymorphic markers in deciphering mixtures. Distinguishing genotypes in a mixed sample. Mixture classification
- •Mixed DNA. Mixture interpretation. Statistical approaches to mixture analysis. chromosomal abnormalities. Issues with low copy number DNA testing. Precautions to ensure optimal results with low-level DNA

PROCTORING: Present. Proctoring ensures the integrity of the exam by monitoring students through cameras and observing their work environment.

Exam Schedule: According to the Timetable. Exam Duration: 180 minutes for written answers to three questions.

Exam Scoring Criteria:

- Theoretical Question 1: Maximum 33 points.
- Theoretical Question 2: Maximum 33 points.
- Practical Question 3: Maximum 34 points.

Exam Grading Scale:

A (90-100%) - the student carefully studied the study material; gives consistent and complete answers to the questions; freely applies the acquired knowledge in practice.

B (75-89%) - the student knows the study material; does not make serious mistakes when answering; can apply the acquired knowledge in practice.

C (60-74%) - the student knows only the basic material, does not always give clear and complete answers.

D (50-59%) - the student has his own ideas about the studied material; cannot fully and correctly answer the questions, makes gross mistakes when answering.

References:

- 1. Burkhard Madea (Editor).Handbook of Forensic Medicine//ISBN: 978-1-118-57062-3. Wiley-Blackwell, 2014. 1312 pages
- 2. Di Maio VJ, Di Maio D. Forensic Pathology. CRC Press New York.

- 3. Gordon I, Shapiro HA, Berson SD. Forensic Medicine A Guide to Principle. Churchill Livingstone New York.
- 4. Spitz WU, Fisher RS. Medico-legal Investigation of Death. Charles Thomas Publishers.
- 5. Norah Rudin, Keith Inman. An introduction to Forensic DNA Analysis. CRC Press, London.
- 6. Robertson J, Ross AM, Burgoyne LA. DNA in Forensic Science Theory, Technique and Application. Ellis Horwood, UK

Professional scientific databases

- 1. https://www.ncbi.nlm.nih.gov/pubmed
- $2\ .\ https://clarivate.com/products/scientific-and-academic-research/research/discovery-and-workflow-solutions/webofscience-platform/$

Internet-resources:

http://elibrary.kaznu.kz/

https://www.ncbi.nlm.nih.gov/pubmed

http://study.com/academy/subj/science.html

https://www.khanacademy.org

https://www.nature.com/scitable/topics

EVALUATION RUBRIC OF THE FINAL CONTROL Discipline: "104830-Molecular and Forensic-Medical Expertise". Form: traditional written/offline

Point	DESCRIPTORS					
	«Great»	«Good»	«Satisfactory»	«Unsatisfactory»		
Criteria	90-100 points	70-89 points	50-69 points	25-49 points	90-100 points	
1. Knowledge and understanding of the theory and concept of the course The answer contains an exhaustive disclosure of all questions (within the limits of acquired knowledge), detailed argumentation of each conclus and statement, is constructed logically and consistently, supported by examples		The answer contains full, but not exhaustive coverage of all issues, abbreviated argumentation of the main points, allows violation of the logic and sequence of presentation of the material. The answer contains stylistic errors, inaccurate use of terms.	The answer contains incomplete coverage of the issues proposed in the ticket, superficially argues the main points, the presentation is allowed to violate the logic and sequence of presentation of the material, does not give examples.	Incorrect coverage of the issues raised, erroneous argumentation, factual stylistic and logical errors, making an incorrect conclusion	Ignorance of the basic concepts of the discipline. Violation of the Rules of final control	
2. Application of the selected methodology to specific applied problems	Complete fulfillment of the task, a detailed, reasoned answer to the question posed when solving a practical problem.	Partial fulfillment of the task, incomplete in some places reasoned answer to the posed question with incomplete solution of the practical task, incorrect terminology used	The material is presented fragmentarily, with logical violation of logical sequence, factual and semantic inaccuracies are admitted, knowledge of problem solving is used superficially	Irrational method of problem solving or insufficiently thought out answer plan, inability to solve problems, fulfill tasks in a general way, making errors and mistakes exceeding the norm.	Inability to apply knowledge, algorithms to solve problems; inability to make conclusions and generalizations. Violation of the Rules of final control	
3. Assessing and analyzing the applicability of the selected methodology to the proposed practical problem, justification of the obtained result	Consistent, logical and correct justification of scientific provisions and applications of the methodology, literacy, visualization of answers	It is allowed 3-4 inaccuracies in the use of conceptual material, minor errors in generalizations and conclusions, which do not affect the good level of performance of the task	Conclusions on the applicability of substantiated scientific provisions are incorrect and unconvincing, there are stylistic and grammatical errors, and inaccuracies in the processing of results.	The task is completed with gross errors, answers to questions are incomplete, conceptual material and reasoning are poorly used	The task is not completed, there are no answers to the questions, the tools of analysis are not used.	
4. Uniqueness of the text (anti-plagiarism)	Basic technical characteristics of the project work; borrowing, self- citation, citation, originality; Final score taking into account the level of originality of the work established faculty (bac, mag, and dock);	Basic technical characteristics of the written work are met; borrowing, citation, originality; large amount of self-citation which does not significantly affect the significance of project examination project exam work; Giving final score taking into account level of originality of the work established by faculty (bak, mag and dock);	Partial compliance basic technical characteristics of the project work; borrowing, citation, originality; large volume of self-citation. which does not significantly affect the significance of project examination project exam work; Giving final score taking into account level of originality of the work established by faculty (bak, mag and dock); Use in the text generative artificial artificial intelligence (GenAI) should be no more than 20%, but in this case the grade on the exam would be 69% or less.	Complete absence of the student's mandatory concepts of modern natural science, skills on the topic being tested, or a significant part of the The originality of the project work on anti-plagiarism has not reached the established threshold level; The use of generative artificial intelligence (GenAI) in the text of the project more than 20% violates the rules of the exam. (GenAI) more than 20% violates the rules of the exam. exam.	The work is perfect technical override (replacement of symbols, insertion of unnecessary symbols, omissions, etc.). Such works after analyzing the full report shall be canceled; Violation of the Rules conducting final Controls. Use in text of the project generative artificial intelligence (GenAI) of more than 20% violates the rules of the exam.	

Example of calculating the final score of written/oral exams

N ₂	Point	«Great»	«Good»	«Satisfactory»	«Unsatisfactory»	
	Criteria	90-100 points	70-89 points	50-69 points	25-49 points	0-24 points
4.	Criteria 1	100				
5.	Criteria 2		75			
6.	Criteria 3			60		
	Final score	100	75	60		100 + 75 + 60 = 235
						235 / 3 criteria = 78,3 Final score = 78

Formula for calculating the final grade:

Final Evaluation (FE) = (P1+P2+P3)/3 C, where P is the criterion score, C is the total number of criteria. Based on the score obtained in the calculation, we can match the score to the grading scale.

78 points are in the range of 70 points to 89 points, which corresponds to the category "Good" according to the evaluation scale.

Thus, in this calculation, the written (oral) work will be evaluated at 78 points "Good" in accordance with the point-rating letter system of evaluation of learning achievements of students with their translation into the traditional grading scale and ECTS. стемой оценки учета учебных достижений обучающихся с переводом их в традиционную шкалу оценок и ECTS.

$Score-rating\ letter\ system\ of\ evaluation\ of\ accounting\ of\ learning\ achievements\ of\ students\ with\ their\ transfer\ to\ the\ traditional\ scale\ of\ grades\ and\ ECTS$

Letter grade	Numerical equivalent	Points (% content)	Traditional system grade
A	4,0	95-100	Great.
A-	3,67	90-94	
B+	3,33	85-89	Good
В	3,0	80-84	
B-	2,67	75-79	
C+	2,33	70-74	
С	2,0	65-69	Satisfactory
C-	1,67	60-64	
D+	1,33	55-59	
D	1,0	50-54	
FX	0,5	25-49	Unsatisfactory
F	0	0-24	Download