


<p>             ОҢТҮСТІК-ҚАЗАҚСТАН  <b>MEDISINA</b>  <b>AKADEMIASY</b>              «Оңтүстік Қазақстан медицина академиясы» АҚ         </p>		 <p>             SOUTH KAZAKHSTAN  <b>MEDICAL</b>  <b>ACADEMY</b>              АО «Южно-Казахстанская медицинская академия»         </p>
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## Methodological instructions for practical classes


**Discipline: "Genes and Heredity" (Molecular biology)**

**Discipline code: GN 1204**

**EP: 6B10115 "Medicine".**

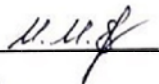
**Volume of study hours/credits: 120 hours/4 credits (12 h)**


**Course and semester of study: 1-2**

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The Methodological instructions for practical classes was developed in accordance with the working curriculum of the EP "Genes and heredity", the discipline "Molecular Biology" and discussed at the meeting of the department.

Protocol no. \_\_18\_\_ of «\_13\_» \_\_06\_\_ 2023 y.

Head of Department, Professor  Yessirkepov M.M.

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**1. Topic:** Structure and functions of proteins and nucleic acids.

**2. Purpose:** Introduction to the structure and function of proteins and the study of the relationship between protein structure and function. To study the structure and function of nucleic acids and sections of the DNA molecule

**3. Learning Objectives:** student should know the structure, function and folding of protein molecule; should be able to identify the structure of protein sequence I, II, III, IV, amino acids; student should know the structure, function, sections of DNA; should be able to identify nucleotides, spatial structure of DNA, sections.

**4. Main topic questions:**

1. Definition of the concept of proteins
2. Structural organization of protein: structure and types of amino acids, definition and essence of peptide bonding
3. Biologically active low molecular weight peptides.
4. Structure of high molecular weight peptides - proteins: primary, secondary, tertiary, super secondary, domain structure of proteins.
5. Quaternary structure of protein.
6. Basic functions of proteins.
7. Functioning of protein. Ligands and their role in the formation of protein structure. Active center.
8. Classification of proteins.
9. The haemoglobin family. Immunoglobulin superfamily. Family of cellular, antigen-recognising receptors. The family of proteins of the major histocompatibility complex. Family of serine proteases.
10. The concept of "export" and internal proteins.
11. p53 protein, its structure and role in the regulation of cellular processes.
12. The role of proteins in nutrition. Changes in protein composition of the organism.
13. Protein folding. Foldases. Chaperones. Refolding.
14. Diseases of disorders of folding. Amyloidoses. Prion diseases.
15. General structure of nucleic acids:
16. Structure of nucleotides,
17. Structure of nucleic acids: linear sequence of nucleotides.
18. Structure of the DNA molecule: Complementarity of nitrogenous bases,
19. Antiparallelism of DNA strands.
20. Nucleosomal strand. Supranucleosomal stacking of DNA.
21. Physical and chemical properties and functions of DNA.
22. Diversity of DNA organisation forms in cells.
23. DNA and protein complexes.
24. Types of RNA involved in translation: mRNA, tRNA, rRNA, tmRNA.


**5. Main forms/methods/techniques of learning** to achieve the final LO of the discipline: Work with microphotographs, diagrams, tables.

**6. Forms of control to assess** the level of achievement of the final LO of the discipline. assessment (testing, solving situational tasks, filling in the medical history, etc.): Testing, oral questioning on CM materials.

**7. Literature:** see appendix 1

**8. Control** (questions, tests, tasks, etc.):

1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

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## №2

**1. Topic:** Molecular mechanisms of genetic information realization. Replication.

**2. Objective:** Explanation of the mechanisms of DNA replication and enzyme complex, participation in preparation for realization and completion of DNA replication.

**3. Learning Objectives:** To study the mechanisms of DNA replication.

**4. Main questions of the topic:**

1. Transfer of genetic information. Three types of transfer of hereditary information. The basic dogma of molecular biology.
2. Reproduction of genetic information. DNA replication: basic principles of DNA replication: conservative, semi-conservative and dispersed replication.
3. three modes of semi-conservative replication: theta-type ( $\theta$ -type), sigma-type ( $\sigma$ -type) and replication of linear molecules.
4. Stages of semi-conservative replication:
  - initiation,
  - elongation,
  - termination.
5. Factors of replication initiation, elongation, and termination.
6. Definition of the concept and function of telomeres.
7. DNA-binding proteins, structure and function.
8. DNA polymerases and their types.
9. PCNA proteins, structure and function.
10. Replication of telomeric sections of DNA.
11. Telomerase. Mechanism of action. Role in the processes of aging and oncogenesis.
12. DNA methylation.

**5. Main forms/methods/techniques of learning** to achieve the final LO of the discipline: Work with microphotographs, diagrams, tables.

**6. Forms of control to assess** the level of achievement of the final LO of the discipline. assessment (testing, solving situational tasks, filling in the medical history, etc.): Testing, oral questioning on CM materials.

**7. Literature:** see appendix 1

**8. Control** (questions, tests, tasks, etc.):

1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

## №3


**1. Topic:** Expression of genetic material. Transcription

**2. Objective:** To give the concept of mechanisms of transcription and translation process; their importance in the chain of biological information transfer in norm and pathology; pathological conditions connected with their violation; mechanisms of their regulation.

**3. Learning objectives:** Study of transcription mechanisms, transcription factors, RNA types.

**4. Main questions of the topic:**

1. DNA transcription is the first stage of expressing information about protein structure.
2. Factors of transcription:
  - common transcription factors;
  - DNA-binding proteins and their types;

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- P-53 protein as a transcription factor.
- 3. Stages of transcription. Initiation, elongation, termination.
- 4. transcription in prokaryotes.
- 5. transcription in eukaryotes.
- 6. inhibitors of transcription.
- 7. precursors of active RNA pre-rRNA, pre-mRNA and pre-tRNA
- 8. pre-rnprocessing. Mechanisms of splicing.
- 9. non-transcriptional addition of individual nucleotides.
- 10. Formation of modifying nucleotides as part of pre-RNA.
- 11. Principles of coding genetic information.
- 12. Genetic code and its properties.
- 13. Main components involved in protein synthesis.
- 14. Aminoacyl-tRNA synthetase.
- 15. Activation of amino acids.
- 16. translation. Occurrence of the incisable complex. Initiation factors.
- 17. Elongation, three steps of translation (elongation of peptide chain). Elongation factors.
- 18. translation termination. Termination factors.
- 19. cytoplasmic and membrane bonding of ribosome. Polyribosomes.
- 20. recruitment of RNA to protein, ribozymes.

**5. Main forms/methods/techniques of learning** to achieve the final LO of the discipline: Work with microphotographs, diagrams, tables.

**6. Forms of control to assess** the level of achievement of the final LO of the discipline. assessment (testing, solving situational tasks, filling in the medical history, etc.): Testing, oral questioning on CM materials.

**7. Literature:** see appendix 1

**8. Control** (questions, tests, tasks, etc.):

1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

#### **№4**


**1. Topic:** Protein biosynthesis. Translation.

**2. Objective:** To familiarize with the structure and function of proteins and to study the relationship between protein structure and function.

**3. Learning objectives:** Study of mechanisms of translation, factors, types of RNA

**4. Main questions of the topic:**

1. Factors necessary for the intensive passage of translation.
2. Principles of coding of genetic information.
3. Genetic code and its properties.
4. Basic components involved in protein synthesis.
5. Aminoacyl-tRNA synthetase.
6. Activation of amino acids.
7. Translation. Emergence of the incisable complex. Initiation factors.
8. Elongation, three steps of translation (elongation of peptide chain). Elongation factors.
9. Termination of translation. Termination factors.
10. Cytoplasmic and membrane communication of ribosome. Polyribosomes.
11. Recruitment of RNA to protein. cjarнк, ribozymes.

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12. The fundamental difference between RNA synthesis and DNA synthesis in the overall structure of the Prokaryotic operon.

13. Lactose operon is an example of an inducible operon.

14. Tryptophan operon is an example of a repressible operon. Regulation with an operon and an attenuator.

15. Catabolite repression.

16. Regulatory mechanism of gene expression in Eukaryotes.

**5. Main forms/methods/techniques of learning** to achieve the final LO of the discipline: Work with microphotographs, diagrams, tables.

**6. Forms of control to assess** the level of achievement of the final LO of the discipline. assessment (testing, solving situational tasks, filling in the medical history, etc.): Testing, oral questioning on CM materials.

**7. Literature:** see appendix 1

**8. Control** (questions, tests, tasks, etc.):

1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

## №5

**1. Topic:** Regulation of gene expression in prokaryotes and eukaryotes.

**2. Objective:** Mechanisms of translation and explanation of the operon theory of gene expression by Jacob and Monod

**3. Learning Objectives:** Induced and repressive regulation of gene expression. Jacob and Monod operon theory. Study of the lactose and tryptophan operon

**4. Main questions of the topic:**

1. Transcription, basic mechanisms and factors
2. Jacob's and monod operon theory
3. translation, basic mechanisms and factors
4. regulation of expression of Repressible Operons
5. regulation of expression of inducible operons
6. lactose and tryptophan operon


**5. Main forms/methods/techniques of learning** to achieve the final LO of the discipline: Work with microphotographs, diagrams, tables.

**6. Forms of control to assess** the level of achievement of the final LO of the discipline. assessment (testing, solving situational tasks, filling in the medical history, etc.): Testing, oral questioning on CM materials.

**7. Literature:** see appendix 1

**8. Control** (questions, tests, tasks, etc.):

1. Answers to test questions.
2. Solving situational tasks.
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## **№6**

**1. Topic:** The hereditary apparatus of the cell.

**2. Objective:** Study of the structure of genetic information, acquaintance with the concept of genome; study of the structure of human genome; study of chromosomal level of genome organisation, morphology of human chromosome, human karyotype.

**3. Learning objectives:** The student should know the structure of genes, types of genes, classification of genes, genome of prokaryotes and eukaryotes; be able to distinguish between the genome of eukaryotes, prokaryotes, viruses, mitochondria.

**4. Main questions of the topic:**

1. Gene is the elementary unit of heredity.
2. Fine structure of gene (exons, introns, cistrons, Mutons, recons.).
3. Classification of genes.
4. Structure of Eukaryotic genes: coding site and non-coding site of eukaryotic genes.
5. Histone gene cluster, ribosomal RNA, haemoglobins.
6. operon structure of prokaryotic genes.
7. give an introduction to the genome.
8. sections of DNA.
9. give an introduction to genetic elements.
10. Explanation of DNA polymorphisms, types (single nucleotide polymorphism, PDRF and vntr).
11. characterisation of DNA fractions.
12. chromosomal complexity of a Alsoobjective DNA.
13. simple tandem repeat (satellites )
14. tandem organisation of clustered genes.
15. genome of cytoplasmic DNA.
16. genome of viruses, bacteria.
17. histones in Chromosome and DNA organisation;
18. Metaphase chromosome;
19. Chromosomatypes:
  - metacentric;
  - submetacentric;
  - acrocentric;
20. function of chromosomes;
21. understanding of karyotype;
22. classification of Karyotypes:
  - Denver;
  - Paris.

**5. Main forms/methods/techniques of learning** to achieve the final LO of the discipline: Work with microphotographs, diagrams, tables.

**6. Forms of control to assess** the level of achievement of the final LO of the discipline. assessment (testing, solving situational tasks, filling in the medical history, etc.): Testing, oral questioning on CM materials.


**7. Literature:** see appendix 1

**8. Control** (questions, tests, tasks, etc.):

1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

## **№7**



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**1. Topic:** Disorders of genetic homeostasis.


**2. Objective:** To explain the exchange of disorders of genetic homeostasis, mutations, biological antimutation in the cell.

**3. Learning objectives:** Study of types of chromosomal mutations and their role in human pathology; the student needs to know the role of mutagenesis in the occurrence of disease, the classification of mutations, the characteristics of mutagenic factors and DNA repair processes; it is necessary to identify the types of mutations and mutagenesis, be able to describe the repair process.

**4. Main questions of the topic:**

1. The concept of homeostasis? Genetic homeostasis.
2. Mutations. Definition.
3. Classification of gene mutations according to their occurrence:
  - exchange of basic pairs
4. Localisation of gene mutations:
  - neutral
  - missense mutation
  - erunda mutation
  - regulator
  - dynamic
5. Genes and classified location of gene mutations in cells.
6. Classification of gene mutations, effect on vital activity of organism: lethal and sub-lethal.
7. Pathological effects of mutations: parental dissociation, imprinting.
8. Single-nucleus polymorphism and its significance in medicine.
9. The concept of chromosomal mutation, or aberration;
10. Classification of chromosomal mutations:
  - a. - Intrachromosomal (deletion; duplication; inversion, hyphenation, ring chromosome, isochromosome);
  - b. - Interchromosomal (translocations, Robertsonian rearrangements).
11. Syndromes arising from chromosomal mutations.
12. Genomic mutations and their types:
  - aneuploidy;
  - polyploidy;
13. Mechanism of genomic mutations;
14. Chromosomal syndromes associated with aneuploidy:
  - Shereshevsky-Turner syndrome;
  - Klinefelter syndrome;
  - Patau syndrome;
  - Edwards syndrome;
  - Down syndrome;
  - X-triplet syndrome.
15. Incidence of chromosomal mutations;
16. Mutagenesis and its types:
  - spontaneous;
  - induction (artificial);
17. Mutagenic factors:
  - physical
  - chemical
  - biological
18. Damage to the DNA molecule:



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19. Damage types of repair in DNA:

- dark;
- light;

20. Biological antimutagenic cell barrier:

- pair of chromosomes;
- DNA repair;
- The matrix nature of DNA synthesis.

**5. Main forms/methods/techniques of learning** to achieve the final LO of the discipline: Work with microphotographs, diagrams, tables.

**6. Forms of control to assess** the level of achievement of the final LO of the discipline. assessment (testing, solving situational tasks, filling in the medical history, etc.): Testing, oral questioning on CM materials.

**7. Literature:** see appendix 1

**8. Control** (questions, tests, tasks, etc.):

1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

## **№8**

**1. Topic:** Molecular-genetic methods of genome research.

**2. Objective:** to form the concept of gene detection, sequencing, modern molecular-genetic methods of human genome research.

**3. Learning objectives:** familiarization with modern molecular-genetic methods of human genome research.

**4. Main questions of the topic:**

1. Give a characterization of DNA diagnostic methods.
  - Sequencing;
  - DNA hybridization;
  - PCR (polymerase chain reaction)
  - \* isolation of the gene under study (DNA fragment);
  - \* Cleavage of DNA chains by heating (annealing);
  - \* Alignment of primers;
  - \* DNA synthesis;
2. Gene detection (use of microarrays).
3. Genetic engineering technologies and their application in medicine.
4. cloning.
5. Transgenic organisms - problems.

**5. Main forms/methods/techniques of learning** to achieve the final LO of the discipline: Work with microphotographs, diagrams, tables.


**6. Forms of control to assess** the level of achievement of the final LO of the discipline. assessment (testing, solving situational tasks, filling in the medical history, etc.): Testing, oral questioning on CM materials.

**7. Literature:** see appendix 1

## **Appendix 1**

**In Russian:**

**Basic:**

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1. Genetics. Textbook for Higher Education Institutions / Edited by Academician of RAMS V.I. Ivanov - Moscow: ICC "Akademkniga", 2006-638c: ill.

2. Muminov T. Fundamentals of molecular biology: a course of lectures. -Almaty: Effekt, 2007.

**Additional:**

1. Ivanyushkin A.Y., Ignatiev V.N., Korotkikh R.V., Siluyanova I.V. Izd-voor Progress, M.. 2008.

2. Y. Clague, M. Cummings. Fundamentals of Genetics - M.: Technosphere, 2009.

3. Fundamentals of molecular biology of the cell. Textbook. 3 volumes. B. Alberts et al, OZON.RU Publishing House, 2018.

№	Name	Link
1	Electronic library	<a href="http://lib.ukma.kz">http://lib.ukma.kz</a>
2	Republican interuniversity electronic library	<a href="http://rmebrk.kz/">http://rmebrk.kz/</a>
3	Electronic library of the Medical University "Student Advisor"	<a href="http://www.studmedlib.ru">http://www.studmedlib.ru</a>
4	"Paragraph" information system "Medicine" section	<a href="https://online.zakon.kz/Medicine">https://online.zakon.kz/Medicine</a>
5	Scientific electronic library	<a href="https://elibrary.ru/">https://elibrary.ru/</a>
6	Electronic library "BuxMed"	<a href="http://www.booksmed.com">http://www.booksmed.com</a>
7	«Web of science» (Thomson Reuters)	<a href="http://apps.webofknowledge.com">http://apps.webofknowledge.com</a>
8	«Science Direct» (Elsevier)	<a href="https://www.sciencedirect.com">https://www.sciencedirect.com</a>
9	«Scopus» (Elsevier)	<a href="http://www.scopus.com">www.scopus.com</a>
10	PubMed	<a href="https://www.ncbi.nlm.nih.gov/pubmed">https://www.ncbi.nlm.nih.gov/pubmed</a>

**Internet resource:**

1. Genetics. Textbook for Higher Education Institutions / Edited by Academician of Russian Academy of Medical Sciences V.I. Ivanov - Moscow: ICC "Akademkniga", 2011-638.

2. Mushkambarov N.N., Kuznetsov S.N. Molecular biology. Textbook for students of medical universities, 3rd edition, Moscow: Nauka, 2016, 660.

3. Y. Clague, M. Cummings. Fundamentals of genetics - M.: Technosphere, 2009.

4. Kurchanov.A. Human genetics with the basics of general genetics: textbook -SPb, 2009.

5. Alberts B. B., Bray D., Hopkin K. Fundamentals of molecular biology of the cell. Textbook. 2nd ed., revised, per. from Engl. 768p. 2018y.

6. Spirin A.S. Protein biosynthesis, the RNA World and the origin of life.

7. Spirin A.S. Molecular Biology. Structure of ribosomes and protein biosynthesis. - M.: (electronic textbook).

**8. Control (questions, tests, tasks, etc.):**

1. Answers to test questions.

2. Solving situational tasks.

3. Filling in cards on the topic.

4. Answering oral questions.