Questions of the Midterm program 1

Title of the discipline: "Molecular Biology and medical genetics"
Discipline code: MBMG 1203
OP name: 6B10115 "Medicine"
Amount of study hours/credits: 180/6
Course and semester: 1-1

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Questions for Midterm No. 1 Molecular Biology

1. Protein monomers. Primary, secondary, and tertiary cell structures. Folding, folding factors. Foldases and chaperones. Classification of proteins and their functions.
2. NK monomers. Nucleoside mono-, di- and triphosphates, formation of a 3,5-phosphodiester bond. The spatial model of DNA. Primary, secondary, and tertiary structures of DNA and RNA. Primary structure of mRNA, secondary structure of tRNA. Ribosomal RNA.
3. Three types of transfer of hereditary information. The basic dogma of molecular biology. DNA replication: basic principles. Replication methods. Replication stages: initiation, elongation, and termination. The composition of the replicome. Factors of initiation, elongation, and termination of replication. Definition of the concept and function of telomeres. Replication of telomeric DNA sections. Telomerase. Role in the processes of aging and oncogenesis
4. Biosynthesis of nucleic acids. Gene expression: DNA transcription. Stages and mechanisms. Transcription factors: DNA and RNA polymerases, general and specific factors. Gna-RNA processing. RNA processing in eukaryotes. Splicing. Informosomes, spliceosomes.
5. Protein biosynthesis. The genetic code and its properties. Types of RNA. Ribosomes. Functional centers of ribosomes. Stages of protein biosynthesis: initiation, elongation, and termination. Protein modification.
6. Regulation of gene activity in prokaryotes at the transcription level: by the mechanism of induction (lactose operon) and repression (tryptophan operon). Regulation of gene activity in eukaryotes: at the level of DNA, transcription, mRNA processing, translation, posttranslation regulation
7. The genetic apparatus of the cell. The gene, its classification, fine structure and properties. The structure of the genes of prokaryotes and eukaryotes. Cluster genes. The genome, DNA departments, and the organization of the human genome. Chromosomes, morphology, classification. Human karyotype. Karyotype classifications
8. The cell cycle and its periods. Cell cycle regulation: cyclins and cyclin-dependent kinases (CCK), mitosis-stimulating factor (MCF). Control points of the cell cycle. The regulatory role of p-53 proteins. Apoptosis.
9. Molecular genetic methods of genome research and their medical significance. Sequencing, DNA hybridization, PCR, and gene detection. Restriction enzymes. Vectors. Plasmids. Cloning without host cells is a PCR method. Molecular cloning. The FISH method
10. Cell membrane: glycocalyx, biomembrane, submembrane layer of musculoskeletal structures. Structure and functions. Lipids of membranes. Types, properties and functions of Micelles and liposomes.Proteins of membranes. Types and functions.
Cytoplasm. Structure and chemical composition.
11. The molecular structure and functions of cellular membrane organelles: The nucleus, the nuclear apparatus of the cell, the structural organization of chromatin, karyoplasm. EPS,

synthesis of EPS proteins. The Golgi apparatus, a three-dimensional model of the Golgi complex. Mitochondria, structural features of the outer and inner membranes. The ATP synthase complex. Lysosomes, classification of lysosomes. Heterophagy, autophagy, and autolysis. Pyroxisomes.

12. Molecular structure and functions of cellular non-branded organelles: ribosomes, cytoskeleton of the cell (microtubules, interstitial and actin filaments). Cytoskeletal proteins. The cellular center. Cilia and flagella. A general idea of the mechanisms of transport of substances.

13. Membrane transport: passive and active.

The direction of membrane transport. The structure and operation of ion channels, translocases, and pumps.

14. Definition and essence of the vesicular transport process. Vesicles. Three ways of vesicle formation. Endocytosis: pinocytosis, phagocytosis, and receptor-mediated endocytosis. Exocytosis: secretion, excretion: recreation: Transcytosis.

15. Intercellular interactions: adhesion, contacts. Families of adhesive membrane proteins. Contacts. Types of contacts: simple type contacts: adhesive and interdigitation; interlocking type contacts - desmosomes and adhesive belts; locking type contacts-tight connection
communication contacts – nexuses and synapses,
matrix-cell contacts: half–desmosomes and focal contacts

Questions for Midterm No. 1 Medical Genetics

1. Fundamentals of classical genetics. Patterns of discrete inheritance of traits. Formulation, characteristics and concepts of the first, second and third Mendelian laws. Solving genetic problems based on Mendelian laws.
Drawing up a Pennet grid. Record the task conditions.

2. Patterns of discrete inheritance of traits. Solving genetic problems based on Mendelian laws.Drawing up a Pennet grid. Recording task conditions

3. Definition of the concept of genotype and phenotype. The interaction of allelic genes: dominance, codominance, overdominance, incomplete dominance.Interaction of non-allelic genes: epistasis: polymerization, its role in the formation of quantitative traits; complementarity. Solving genetic problems based on Mendelian laws.
Drawing up a Pennet grid. Recording task conditions

4. Definition of the concept of inheritance linked to gender. Characteristics of inheritance types. Solving problems of linked inheritance and gender-linked inheritance

5. Definition of the concept of inheritance linked to gender. Characteristics of inheritance types. Solving problems of linked inheritance and gender-linked inheritance

6. Definition of the concept of genetic homeostasis. Violation of homeostasis is the cause of diseases. Mutations. Definition. Classification of gene mutations. Classifications of

chromosomal and genomic mutations. Biological antimutagenic barriers of the cell. Homogenous dysomias, imprinting. Single nucleoid polymorphism. The concept of chromosomal aberrations; Mutagenesis and species. Mutagenic factors.Definition of concepts. Phases of fertilization. Conditions of fertilization. The gamoons. Capacitation, Monospermia. Acrosome and cortical reactions. Dicarion, Syncarion. The zygote. Parthenogenesis. Gynogenesis and androgenesis.

7. Violation of genetic homeostasis. Definition of the concept of chromosomal and genomic mutations. Mechanisms of development of chromosomal and genomic mutations. Classification of mutations: intra-chromosomal and inter-chromosomal, aneuploidy, Polyploidy.

8. Definition of the concept of environment and environmental factors.

The reaction rate. Forms of reaction norms. Definition of the concept of genocopy and phenocopy. Definition of the concept of variability. Types of variability: genotypic, phenotypic, modification and random, combination and mutation. Generative and somatic variability

9. The subject and objectives of medical genetics.

Features of the study of human genetics. Methods of studying human genetics: twin, dermatoglyphics and palascopy, genetics of somatic cells, popularly static, biochemical, cytogenetic, clinical and genealogical.

10. Diseases with a non-Mendelian type of inheritance: mitochondrial, genomic imprinting; expansion of trinucleotide repeats.

11. Hereditary diseases. Genetic mechanisms of occurrence.Monogenic diseases. Classification of monogenic diseases. Polygenic (multi-factorial) diseases (MB). General characteristics and classification of MB. Approaches to the study of hereditary predisposition to human diseases. Molecular genetic analysis of the mechanisms of MB development.

12. Chromosomal diseases.

Classification of chromosomal diseases:

Multiple congenital malformations (MVP). Etiology, clinic and genetics of syndromes.

13. Diseases with a non-Mendelian type of inheritance: mitochondrial, genomic imprinting; expansion of trinucleotide repeats.

14. The genetic basis of the prevention of hereditary diseases. Prenatal diagnosis. Preimplantation diagnostics of the basics of human ecology. Definition of the concept of biotransformation. Oxidative stress. Pharmacogenetics. The effect of medicinal preparations on the human hereditary apparatus. Definition of the concept of predictive medicine. Genetic foundations (genetic certification), prospects, medical significance.

15. Definition of the concept of population genetics. Population structure. The gene pool. The genetic unity of the population is panmixia. The genetic unity of the population. The Hardy-Weinberg Law. Elementary evolutionary factors: mutations, gene migration, gene drift, natural selection, population waves.

Questions for Midterm No. 1 Embryology

1. Definition of the concepts of developmental biology and ontogenesis. Basic concepts in the biology of individual development. Classifications of ontogenesis. Stages of ontogenesis
2. Definition of the concept of a cell. Cytoplasm. Cytolemma, Simplast, syncytium. Organelles. Definition, classification, structure, and functions of membrane-type and non-membrane-type organelles. Classification of inclusions. Types and structure.
3. Mitosis and meiosis. Phases. Features and differences. Atypical mitosis.
4. Definition of the concept. Gonocytes. Zones of gametogenesis. The essence of the process. Spermatogenesis and oogenesis. Ovogenesis: localization, stages, age-related changes. The construction of a mature human egg. Classification of eggs.Spermatogenesis: localization, stages, age-related changes. The structure and number of spermatozoa. Male fertility. The main events occurring in human ovogenesis and spermatogenesis.
5. Egg cell: structure, types according to the amount of yolk and its location, shell.
Spermatozoon: the head of the spermatozoon, the acrosome and the tail section
6. Definition of concepts. Phases of fertilization. Conditions of fertilization. The gamoons. Capacitation, Monospermia. Acrosome and cortical reactions. Dicarion, Syncarion. The zygote. Parthenogenesis. Gynogenesis and androgenesis.
7. Fragmentation and its types; the difference between embryo fragmentation and mitotic division of somatic cells; structure and types of blastules; blastocyst.
8. Definition of the concept, structure of gastrula; methods of gastrula formation. Differentiation of the mesoderm. Formation of axial and pharmacological organs. Germ layers and their derivatives. Mesoderm and mesenchyme, their derivatives.
9. Definition of the concept. Differentiation of ectoderm, endoderm. Methods of mesoderm formation. Mesenchyme. Their derivatives
10. The genealogical method, symbols, the principle of compiling pedigrees and its analysis. Calculation of the genetic risk of the disease
11. Definition of concepts. Cellular mechanisms underlying the processes of ontogenesis. The concept of determination. Differentiation of cells during embryogenesis. The molecular genetic basis of differentiation. Potency: totipotence, pluripotence, multipotence,
Unipotence. Integration in ontogenesis. The concept of embryonic induction. Proliferation is the multiplication of cells. Cell migration. Sorting and clumping of cells (adhesion). Programmed cell destruction during embryogenesis is apoptosis. Types of programmed cell death. The role of apoptosis in morphogenesis. Polypotence (totipotence), unipotency, and cell determination
12. Definition of the concept. The structure, topography, and functions of the extra-embryonic organs of the human embryo. amnion, yolk sac, allantois, chorion, placenta. The fetal part of the human placenta. Sources of extra-germ organ tissue development
13. Periods of development: initial, embryonic and fetal. Features of fertilization, implantation, cleavage, and blastocyst. Fragmentation, neurulation. Formation of axial organs. Separation of the embryo body. The timing of embryo development. Growth and development

14. Ontogenesis and genes of the maternal organism are fundamental patterns of development and growth based on gene influences. The genetic basis of differentiation. Genes are morphogens. Segmentation genes. Homeotic genes

15. Causes of development and classification of congenital malformations. Pathogenesis of congenital malformations. Critical periods. Diagnostic methods of VPR. Prevention of congenital malformations. development. Definition of the concept. Dates of the event. Methods of prenatal diagnosis of genetic and congenital diseases. Non-invasive (without surgical intervention) methods of prenatal diagnosis: ultrasound, X-ray (feto-amneography).Invasive methods of prenatal diagnosis: amniocentesis, choriocentesis (chorionic biopsy), cardiocentesis (vascular puncture) of the umbilical cord of the fetus. Indirect methods: investigation of the concentration of alpha-fetoprotein (ACE), estriol, gonadotropin in the blood serum of a pregnant woman.