Methodical instructions for independent work of students

Module: "Genes and heredity"

Discipline: Medical Genetics

Code of discipline: GN 1204

Name of EP: 6B10115 "Medicine"

Course hours/ credit hours: 120 hours/4 credits

Course and semester of study:1-2

Shymkent 2023

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

Protocol no. __6_ of «_28__» _12_____ 2023 y.

Head of Department, Professor _____ Yessirkepov M.M.

№ 1

1. Topic: Human Karyotype. Genetic maps of human chromosomes. Anatomy of normal and pathological karyotype

2. Purpose: study of karyotype, its classification, method of mapping genes of genes to obtain gene position and determination of linkage groups.

3. Tasks:

1. Define the concept of genetic map of chromosomes
2. G. Sturtevant and the first genetic map of Drosophila chromosomes
3. Stages of gene mapping
4. Cytogenetic maps
5. Linkage groups and their determination by mapping
6. Genome maps
7. Anatomy of the normal karyotype.
8. Anatomy of pathological karyotype.

4. Form of performance/assessment: Work in small groups, defense of the presentation, glossary.

5. Criteria and fulfillment: Oral questioning.

6. Due date: 1 week

7. Literature: see appendix 1

8. Control:

1. Answers to test questions.
2. Solving situational tasks.
3. Completion of cards on the topic.
4. Answers to the questions indicated in the assignments.

Optional: oral answers to questions on the topic.

№ 2

1. **Topic:** Hereditary apparatus of cells. Dynamics of hereditary apparatus during cell cycle process

2. Objective: to study the structure of the genetic apparatus of the cell at the chromosomal level of genome organization; to study the dynamics of the hereditary apparatus during the cell cycle.

3. Learning objectives: the student should know the structure of the hereditary apparatus at the chromosomal level; be able to describe the changes of the hereditary apparatus during the cell cycle.

4. Tasks:

1. Definition of the concept of hereditary material
2. Structural organisation of chromatin
3. The problem of compactisation of the DNA molecule
4. Levels of DNA molecule compactisation:

-nucleosomal strand chromatin fibril, chromomers and chromonemes, euchromatin and heterochromatin

5. Polytene chromosomes.
6. Lampbrush type chromosomes

4. Form of performance/assessment: Small group work, defense of presentation, glossary.

5. Criteria and fulfillment: Oral examination.

6. Due date: 1 week

7. Literature: see appendix 1

8. Control:

1.Answers to test questions.
2.Solving situational tasks.
3.Completion of cards on the topic.

4.Answers to the questions indicated in the assignments.
Optional: oral answers to questions on the topic.

№ 3
1. Topic: Molecular structure of cells and diseases arising from their malfunctioning
2. Purpose: On the basis of the study of plant and animal cells to show the unity of organization of living forms on our planet. To know the difference between plant and animal cells. To get acquainted with the molecular structure and functions of the cell
3. Tasks:

1. Molecular structure and functions of the major components of the cell:
2. Diseases associated with abnormalities of the nucleus:
3. reduction of genetic material
4. atypical mitoses
5. pathology of synthesis of ribosome subunits and tRNA in the nucleus
6. 2 Diseases associated with disorder of EPS functioning and structure:
7. EPS cisternae dilatation, EPS fragmentation, PS hyper- and hypotrophy, blockade of synthetic and/or transport processes in the cell.
8. diseases associated with disorder of functioning and structure of the Golgi apparatus:
9. Diseases associated with disruption of intracellular transport signals
10. Diseases associated with impaired mitochondrial function and structure:
11. mitochondrial diseases associated with defects in nuclear DNA
12. mitochondrial diseases caused by mtDNA defects
13. Diseases associated with disorders of lysosome function and structure:
14. Mucopolysaccharide accumulation diseases or genetic accumulation diseases;
15. Diseases associated with disorders of sorting and transport of lysosomal enzymes - hydrolases.
16. Diseases associated with damage to lysosomal membranes.
17. diseases associated with extracellular release
18. The role of lysosomes in the development of inflammatory processes
19. Diseases associated with disruption of peroxisome function and structure:
20. Diseases resulting from near total loss of peroxisomal function;
21. Diseases arising due to excess of peroxisomal enzymes;
22. Diseases associated with membrane dysfunction.
23. Diseases associated with changes in the structure and number of cytoskeleton elements.

4. Form of performance/assessment: Small group work, defense of the presentation, compilation of a glossary.
5. Criteria and performance: Oral questioning
6. Due Date: Week 5
7. Literature: see appendix 1
8. Control:
1. Answers to test questions.
2. Solving situational tasks.
3. Completion of cards on the topic.
4. Answers to the questions indicated in the assignments.
Optional: oral answers to questions on the topic.

№4
1. Topic: Modern classification of mutations and examples of inherited diseases

2. Purpose: to familiarize with the classification of mutations and examples of hereditary diseases caused by them

3. Tasks:

1. Definition of the concept of mutations.
2. Classification of mutations:

By place of origin:
a. generative (in germ cells)
b. somatic (in somatic cells)
by adaptive significance:
a. harmful (lethal and semi-lethal)
b. beneficial
by nature of manifestation:
a. dominant
b. recessive.
by the direction of gene change:
a. direct
b. reverse
by the nature of the change in the genotype:
a. genetic,
b. chromosomal
c. genomic
3. Classification of gene mutations:
a. by mechanism of occurrence
b. by the nature of the consequences
4. Classification of chromosomal mutations:
a. by the nature of structural changes
b. by the nature of quantitative changes.
5. Examples of diseases caused by these mutations

4. Form of performance/assessment: Small group work, defense of the presentation, compilation of a glossary.
5. Criteria and performance: Oral questioning
6. Due Date: Week 5
7. Literature: see appendix 1
8. Control:
1. Answers to test questions.
2. Solving situational tasks.
3. Completion of cards on the topic.
4. Answers to the questions indicated in the assignments.
Optional: oral answers to questions on the topic.

№5

1. Topic: Monogenic diseases arising due to changes in protein structure
2. Purpose: Description of molecular-genetic mechanisms of pathogenesis of monogenic diseases.
3. Tasks:
1. Introduction
2. State of protein metabolism in norm
3. Change of protein balance in the process of individual development
4. Changes in protein metabolism in pathological conditions.

4. Form of performance/assessment: Work in small groups, defence of the presentation, compilation of glossary.
5. Criteria and fulfillment: Oral questioning.
6. Due date: Week 11
7. Literature: see appendix 1
8. Control:
1. Answers to test questions.
2. Solving situational tasks.
3. Completion of cards on the topic.
4. Answers to the questions indicated in the assignments.
Optional: oral answers to questions on the topic.

№6
1. Topic: Advances in genetic engineering and their use in medicine: gene therapy, translational therapy. Nanobiotechnology.
2. Purpose: To give an idea of gene therapy and translational therapy. To describe the essence of bionanotechnology and their application in medicine
3. Tasks:
1. Recombinant DNA techniques
2. Genetic diseases: diagnosis and screening
3. DNA microarrays and genetic screening
4. Gene therapy, translational therapy and ethical issues.
5. Creation of recombinant DNA
6. Restriction enzymes
7. Cloning in E.coli bacterial cells
8. Cloning without host cells - PCR method
9. Regenerative medicine
10. Tissue engineering
11. nanotechnology, nanobiotechnology and nanomedicine.
12. Definition of the concept and essence of nanobiotechnology.
13. Processes of nano-biotechnology,
14. Basic steps of biotechnological processes.
15. Clinical (pharmaceutical) nanobiotechnology.
16. Successes and achievements of nanobiotechnology.

4.Form of performance/assessment: Work in small groups, defence of the presentation, glossary compilation. 4.
5.Criteria and fulfilment: Oral questioning.
6. Due date: Week 11
7. Literature: see appendix 1
8. Control:
1.Answers to test questions.
2.Solving situational tasks.
3.Completion of cards on the topic.
4.Answers to the questions indicated in the assignments.
Optional: oral answers to questions on the topic.

№7
1. Topic: Prenatal diagnosis and prevention of hereditary diseases

2. Objective: to study the basics of prenatal diagnosis, medical and genetic counselling and modern methods of prevention of hereditary diseases.

3. Tasks:

1. Genetic bases of prevention of hereditary diseases:
- primary prevention
- secondary prevention
- tertiary prevention
- gene expression management
- elimination of embryos and foetuses with hereditary pathologies
- germ cell genetic genetic inheritance t
- family planning
- environmental protection
2. Medical and genetic counselling
3. Prenatal diagnostics:
   - screening of pregnant women based on the determination of biochemical markers
   - invasive methods:
- amniocentesis
- cordocentesis
- chorionic and placentobiopsy
  - non-invasive methods:
- ULTRASOUND
4. Preimplantation diagnostics
5. Preclinical diagnosis, screening programmes and preventive treatment

4.Form of performance/assessment: Work in small groups, defense of the presentation, glossary writing.

5.Criteria and fulfillment: Oral questioning

6. Due date: 13th week

7. Literature: see appendix 1

8. Control:

1.Answers to test questions.
2.Solving situational tasks.
3.Completion of cards on the topic.
4.Answers to the questions indicated in the assignments.

Optional: oral answers to questions on the topic.

№8

1. Topic: Polygenic diseases. Chromosomal diseases

2. Purpose: Study of etiology and pathogenesis of polygenic diseases (diseases with hereditary predisposition - DHP). Study of etiology, pathogenesis and epidemiology of chromosomal diseases.

3. Tasks:

1. General characterization and classification of DHP
2. Approaches to the study of hereditary predisposition to human diseases
3. Molecular-genetic analysis of mechanisms of development of DHP
4. Genes of susceptibility to some multifactorial diseases
5. Clinical and genetic features of some diseases with hereditary predisposition:

a) артериальная гипертензия
c) bronchopulmonary diseases

d) Diabetes mellitus
e) Gastric and duodenal ulcer disease
f) Alzheimer's disease
g) immune-related diseases
h) infectious diseases
i) malignant neoplasms
6. Classification of chromosomal diseases according to the mechanism of their occurrence.
7. Etiology, clinic and genetics of the syndrome caused by X-chromosome monosomia
8. Main features of clinical picture
9. Etiology, clinic and genetics of diseases caused by X-chromosome polysomy in women and men
10. Main features of clinical picture
11. Etiology of diseases caused by Y-chromosome polysomy 11.Etiology of diseases caused by Y-chromosome polysomy
12. Clinic and genetics
13. Etiology, clinic and genetics of syndromes due to monosomy on autosomes.
14. Etiology, clinic and genetics of syndromes caused by polysomy on autosomes.
15. Etiology, clinic and genetics of syndromes caused by partial autosomal monosomy.

4. Form of performance/assessment: Work in small groups, defense of the presentation, glossary compilation
5. Criteria and performance: Oral questioning.
6. Due date: Week 14
7. Literature: see appendix 1
8. Control:
1. Answers to test questions.
2. Solving situational tasks.
3. Completion of cards on the topic.
4. Answers to the questions indicated in the assignments.

Optional: oral answers to questions on the topic.

№9

1. Topic: Midterm control No. 2 on sections: "Fundamentals of Medical Genetics"
2. Purpose: to determine the level of students' mastery of the material of the passed topics on the sections "General and Medical Genetics".
3. Learning objectives: the student should know the material on the given topic; be able to fulfill the tasks of measuring instruments (oral questions, tests, solving situational tasks, working with cards).
4. Main questions of the topic:

1. Definition of the concept of cell and mitotic cycles.
2. Periods of the cell cycle: G1, S, G2, M, G0; processes occurring during these periods.
3. Types of cells with different abilities to divide:
4. - mitotic,
5. - irreversible postmitotic cells
6. -irreversible postmitotic cells.
7. Cell division - mitosis. Biological significance.
8. Atypical mitoses. Causes of occurrence and significance for medicine.
9. Cell cycle regulation: cyclins and cyclin-dependent kinases, their role in mitotic cycle regulation.
10. Mitosis-stimulating factor.
11. Mechanism of action of cyclin-sdk complexes:

12. - In G_1-period;
13. - in S and G_2-periods;
14. - cell cycle checkpoints.
15. Regulatory role of p-53 protein;
16. General idea of the mechanism of apoptosis. Types of apoptosis: "apoptosis from within" and "apoptosis on command".
17. 17. Mitochondrial factors of apoptosis and the role of p-53 protein.
18. 18. Apoptosis and necrosis. The role of apoptosis in the maturation and functioning of the immune system;
19. 19. Definition of the concept of carcinogenesis.
20. 20. Genetic nature of carcinogenesis. Carcinogenic factors.
21. 21. Biological mechanisms of carcinogenesis.
22. 22. Definition of the concept of adhesion.
23. 23. Families of adhesive membrane proteins
24. -integrins;
25. -selectins
26. -adhesive immunoglobulins
27. -cadherins
28. 24. Adhesive function of membranes
29. -mechanism of T-lymphocyte homing
30. -mechanism of T-cell migration
31. -inflammatory response and adhesion
32. -immune reactions
33. 25. Intercellular contacts. Types of contacts:
34. -simple intercellular junctions
35. -interdigitation
36. -adhesive belt
37. -tight junction
38. -nexus or slit-like connection
39. 26. Extracellular matrix.
40. 27. Definition of the concept of cell signalling.
41. 28. Intercellular signalling substances - primary mediators.
42. 29. Membrane bound and intracellular receptors.
43. 30. Secondary mediators.
44. 31. Basic steps in signal transduction. Medical significance.
45. 32. Mechanisms of intracellular vesicular transport of substances
46. - transport of low molecular weight compounds
47. - simple diffusion
48. - facilitated diffusion
49. - active transport
50. 33. Ion channels. Structure and function.
51. 34. Active transport. Translocases.
52. 35. Direction of transport of substances: uniport, symporti antiport.
53. 36. Ion pumps. Structure and functions.
54. 37. Types of pumps:
55. - Na+K+ - pump
56. - Na+ - channels
57. - K+- channels

58. - Cation channels and n-cholinoreceptors
59. Transport of high molecular weight compounds across membranes
- endocytosis
- exocytosis
60. Subject and objectives of medical genetics.
61. Features of the study of human genetics.
62. Methods of studying human genetics.
63. Hereditary diseases. Mechanisms of occurrence. Genetic mechanisms of hereditary diseases.
64. Monogenic diseases. General characteristics of monogenic pathology.
65. Classification of monogenic diseases:
-by type of inheritance:
-by organ and systemic type:
-by etiology:
-by violation of the type of metabolism
66. Polygenic (multifactorial) diseases (MB).
67. Features of polygenic diseases.
68. General characteristics and classification of MBs.
69. Clinical and genetic features of some MBs:
-arterial hypertension
-broncho-pulmonary diseases
-diabetes mellitus
-gastric and duodenal ulcer disease
-Alzheimer's disease
-immune-related diseases
-infectious diseases
-malignant neoplasms
70. Genocopies and phenocopies.
71. Define the concept of congenital malformations (CMD).
72. Genetic mechanisms of embryogenesis, violation of which leads to the occurrence of CHD.
73. Classification and etiology of CHD.
74. Congenital malformations of multifactorial diseases.
75. Chromosomal diseases and their place in general human pathology.
76. Classification of chromosomal diseases:
-ethiological (based on the nature of the mutation):
-Diseases related to the type of cells in which the mutation occurred (in gametes or zygote):
-diseases related to the time of occurrence of the mutation (in a generation):
77. Multiple congenital malformations (MCMDs) characteristic of chromosomal diseases:
78. Etiology, clinic and genetics of syndromes caused by:
- X-chromosome monosomy;
- X-chromosome polysomy in women and men
- Y-chromosome polysomy
- autosomal monosomy.
- autosomal polysomy.
-partial monosomy on autosomes.
79. Definition of the concept of diseases by the Snemendelian type of inheritance.
80. Causes and mechanisms of development:
- mitochondrial diseases;
- genomic imprinting diseases;

- Trinucleotide repeat expansion diseases;
- prion diseases;

81. General characterisation of the principles of treatment of inherited diseases:
82. Regenerative medicine.
83. Tissue engineering.
84. Genetic basis of prevention of hereditary diseases:

- primary prevention
- secondary prevention
- tertiary prevention
- gene expression management
- elimination of embryos and foetuses with hereditary pathologies
- genetic engineering at the germ cell level
- family planning
- environmental protection

85. Medical and genetic counselling
86. Prenatal diagnostics:

- Pregnancy screening based on the determination of
-biochemical markers
- invasive methods:
- amniocentesis
- cordocentesis
- chorionic and placentobiopsy
- non-invasive methods:
- ULTRASOUND

87. Preimplantation diagnostics
88. Pre-clinical diagnostics, screening programmes and preventive treatment
89. Population, definition.
90. Ecological structure of a population.
91. Genetic structure of a population: genetic unity and genetic polymorphism.
92. Genetic unity (Hardy-Weinberg law) of a population. The Hardy-Weinberg Law and its significance for medicine
93. Human population structure, its characterisation and types: Mendelian, demes, isolates.
94. Genetic polymorphism - characteristic of genetic diversity of a population.
95. Types of genetic polymorphism: adaptation and balanced polymorphism
96. Genetic load - source of recessive allele's appearance
97. Geneogeography of hereditary diseases

4. Form of performance/assessment: Testing, solving genetic problems, oral questioning
5. Criteria and performance: Appendix 2.
6. Due date: 14th week
7. Literature:
Appendix 1
In Russian:
Basic:

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., SiluyanovaI.V.Izd-voor Progress, M.. 2008г.

2. У. 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	http://lib.ukma.kz
2	Republican interuniversity electronic library	http://rmebrk.kz/
3	Electronic library of the Medical University "Student Advisor"	http://www.studmedlib.ru
4	"Paragraph" information system "Medicine" section	https://online.zakon.kz/Medicine
5	Scientific electronic library	https://elibrary.ru/
6	Electronic library "BuxMed"	http://www.booksmed.com
7	«Web of science» (Thomson Reuters)	http://apps.webofknowledge.com
8	«Science Direct» (Elsevier)	https://www.sciencedirect.com
9	«Scopus» (Elsevier)	www.scopus.com
10	PubMed	https://www.ncbi.nlm.nih.gov/pubmed

Internet resource:

1. Genetics. Textbook for Higher Education Institutions / Edited by Academician of Russian Academy of Medical Sciences V.I. Ivanov - Moscow: ICC "Akademkniga", 2011-638c: ill.
2. Mushkambarov N.N., Kuznetsov S.N. Molecular biology. Textbook for students of medical universities, 3rd edition, Moscow: Nauka, 2016, 660c.
3. У. 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. 768st. 2018г.
6. Spirin A.S. Protein biosynthesis, the RHK World and the origin of life.
7. Spirin A.S. Molecular Biology. Structure of ribosomes and protein biosynthesis. - M.: (electronic textbook).

8. Control: