**National University named after Al-Faraby**

**Biology and biotechnology department**

**Molecular biology and of genetics division.**

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| --- | --- |
|  | **Biology and biotechnology department**  Scientific council on meeting gained a foothold  №\_\_\_\_protocol " \_\_\_\_"\_\_\_\_\_\_\_\_ 2014 year.  Dean of faculty Shalahmetova T.M. |

**Speciality 5B010700 Biotechnologies**

**Syllabus**

**Instantaneousness**

**Э 16 Medical genetics bases**

**Semester: Autumn, 3 courses**

**Number credits: 3**

**Type of object: necessarily**

Centre organizing public lectures:

Bigaliyev Aitkhozha Bigaliyevich Biology Science Doctor, molecular biology and of genetics division, Supreme school of KR National Academy academician, 8(727) - 377- 33-34 12-15 additions, 8 775-207-46-13, Aitkhozha.Bigaliev@kaznu.kz , aitkhazha@gmail.com:

Object duties and aims:

Aims: what adding to students medical by bases of genetics uses, modernly genetically, diagnostically and to investigate about methods fully to pass notification.

Duty:

- classic bases of genetics conformities to law the use in industry of medicine, and to search modern methods that uses to investigate patients; cytogenetic, to investigate origin, twins method analysis, sex of man to expose (to search chromatin in somatic cells Х and Y Poisons to expose), by using population and statistics method.

Competences (results educating) : in result reading of student object classic bases of genetics conformities to law the use in industry of medicine studying, meets modern by methods, uses that to investigate on diagnostics of patients.

Prerekvisits: general genetics, high molecularness genetics, ecological genetics, biochemistry, chemistry, physics, mathematics, bases of physiology.

Postrekvisits: man genetics, cytogenetics of man, monogenic and multigenis bases of patients, oncologic genetics of patients.

Э 16 Medical genetics bases

National University named after Al-Faraby, molecular biology and of genetics division

Author : Biology Science Doctor, molecular biology and of genetics division, Supreme school of KR National Academy academician, Pr. Bigaliyev A.B.

Working program molecular biology and genetics division of meeting carry out

" " 2014 year. Protocol NO:

Chief of division

Biology Science Doctor, Pr. Z.G. Aitasheva

Department of biology methodically meeting carry out

" " 2014 year. Protocol NO:

Chairman A.B. Goncharova

**OBJECT MAINTENANCE OF STRUCTUR**

|  |  |  |  |
| --- | --- | --- | --- |
| WEEK | Lessons | Number of Hours | Grade |
| 1 Instantaneousness Medical genetics opened, development and modern the state | | | |
| 1 | 1) Lesson. Introduction to medical genetics history (development med-genetics. Objects and aims. | 1 |  |
| 1) practical (laboratory) lesson. Medical genetics research methods. |  |  |
| 1) Quiz |  |  |
| 2 | 2) lesson. In medicine clinic-genealogy investigates qualities of heredity a method. | 1 |  |
| 2) practical methods qualities of heredity Man research (laboratory) lessons population and statistically methods |  |  |
| 2) Quiz |  |  |
| 3 | 3) lessons about chromosomal disease of patients and modern data | 1 |  |
| 3) practical (laboratory) lesson. Х and Y chromatin poison to expose that are related to the sexual by the method of chromatin and Prenatal patients to expose (diagnostics) |  |  |
| 3) Quiz |  |  |
| **2**  Module **Medical bases of genetics. Consult medical-genetics** | | | |
| 4 | 4) lessons Principle of medical-genetics consultation. | 1 |  |
| 4) practical (laboratory) research methods to estimated twins. Genomic and chromosomal changeable methods exposure. |  |  |
| 4) Quiz |  |  |
| 5 | 5) Lesson. Corresponding to conformity to law of Mendel in genetics of man hereditary of patients. | 1 |  |
| 5) practical(laboratory). Monogenic and polygenic patients diseases to discuss (examples). |  |  |
| 5) Quiz |  |  |
| 6 | 6) Lesson. In medical genetics gender mutual the impression, great number on example of allellism. | 1 |  |
| 6) practical (laboratory).Great Number allellism of blood types the carp to drive according to the АВО system of blood and Rh (rhesus-factor). |  |  |
| 6) Quiz |  |  |
| 7 | 7) Lesson. Test, in uses that on medical genetics the state of in vivo, will systematize. | 1 |  |
| 7) practical (laboratory). Micronuclear test. I'ts heredity specifies that the impression.  That was dedicated heredity number values the impression.. |  |  |
| 7)Quiz |  |  |
| Intermediate control (exam) | 30 |  |
| 7) Lesson. Test, in uses that on medical genetics the state of in vivo, will systematize. |  |  |
| **3** Module To evaluation of radiation effect of medical genetics **and research methods** | | | |
| 8 | 8) Lesson. Medicine and radiation mutagenesis.   Small doses irradiated effects and bases of radiobiology. | 1 |  |
| 8) practical (laboratory) lesson. Molecular research methods into radiation mutagenesis and DNA damage, free radicals effect. |  |  |
| 9 | 9) Lesson. Induced by radiation and spontaneous mutations is influence to man developing processing. Genetics of human congenital defects. | 1 |  |
| 9) ) practical (laboratory) lesson. Radiation effects to spontaneous abortions and congenital defects size of human. |  |  |
| 9)Quiz |  |  |
| 10 | 10) lesson. Modern prophylaxis methods of hereditary diseases. | 1 |  |
| 10 practical (laboratory) lesson. To evaluation of genetically consequence of genomic and chromosomal mutations. |  |  |
| 10)Quiz |  |  |
| 11 | 11) Lesson. Role of small dose radiation effects and biological types of reaction of body. | 1 |  |
| 11) practical (laboratory) lesson. To use of biological dosimeter method. |  |  |
| 11)Quiz |  |  |
| 12 | 12) Lesson. Positive results of adaptation and homeosis processing to small dose irradiated effect on body. | 1 |  |
| 12) practical (laboratory) lesson. Phil genetically similar of Mammalian body to in vivo sex cell. |  |  |
| 12)Quiz |  |  |
| 13 | 13) Lesson. Test-systems, that uses of medical genetics and their classification. | 1 |  |
| 13) practical (laboratory) lesson. Review  of test-systems at modern medical genetics.  Computer systems and information bases (OMD, POSSUM). |  |  |
| 13) Quiz |  |  |
| 14 | 14)lesson. Gene mutations, gene amplification, chromosome instability and reparation processing. | 1 |  |
| 14) practical (laboratory) lesson. Patients with genic disease and to investigate of human hereditary by biochemical methods. |  |  |
| 14) Quiz |  |  |
| 15 | 15) Lesson. Radiation and mutagenic safety. | 1 |  |
| 15) practical (laboratory) lesson. Structure of the test-systems to esteemed radiation safety and request is to test-systems. |  |  |
| 15) Quiz |  |  |
|  |  |  |
| **Intermediate control (midterm)** | **30** |  |
|  | **Exam** | **40** |  |
|  | **Total** | **100** |  |

LITERATURES  LIST

Basic:  
1. Бочков Н.П., Боде А. Медицинская генетика, учебник, М., Наука, 2002   
2. Гинтер А.К Медицинская генетика. Учебник, М.,Medicine, 2003  
3. Сойфер В.Н. Исследования геномов к концу on 1999года // Соросовский образовательный a magazine. 2000. №6. С.15-22.   
4.Глик Б., Пастернак Дж. Молекулярная биотехнология.Принципы bend применение. М., Мир. 2002.  
6.Watson J.D. (2004) DNA. The Secret of Life. Arrow Books.London  
  
is Addition:  
1.Горбунова В.Н. Медицинская генетика. Учебник для студентов слушателей послевузовского образования. М., Medicine, 2010   
2.Biology человека. Чебышев В.Г. и др., М., Высшая школа. 2009  
3.Медицинская генетика, учебно-методическое пособиедля студентов,  
врачей-интернов, ординаторов, педиатров.Благовещенск

4. Клиническая диагностика врожденных пороковразвития. Методическое пособие для студентовмедицинских ВУЗов bend врачей. - М.: ГОУ ВУНМЦ МЗРФ, 2001. - 32 с.  
5.Козлова С.И. с соавт. "Наследственные синдромы bendмедико-генетическое консультирование".М, 1996 г.  
6.Wolker Sh. Biotechnology /The McGraw - Hill Companies. 2007. - 336 P.

**Policy of academic conduct and ethics**

Be tolerant and respect other people's opinions. Objections formulated in the correct form. Plagiarism and other forms of cheating are not allowed. Prompting unacceptable and cheating during delivery SSW interim control and examination, copying solved tasks others exam for another student. A student convicted of falsifying any information of the course will receive a final grade «F».

**Scale of assessment of knowledge:**

|  |  |  |  |
| --- | --- | --- | --- |
| Score on alphabetic system | Digital equivalent | % | Score on classical system |
| А | 4,0 | 95-100 | Excellent |
| А- | 3,67 | 90-94 |
| В+ | 3,33 | 85-89 | Good |
| В | 3,0 | 80-84 |
| В- | 2,67 | 75-79 |
| С+ | 2,33 | 70-74 | satisfactorily |
| С | 2,0 | 65-69 |
| С- | 1,67 | 60-64 |
| D+ | 1,33 | 55-59 |
| D- | 1,0 | 50-54 |
| F | 0 | 0-49 | unsatisfactorily |
| I  (Incomplete) | - | - | Incomplete |
| P  (Pass) | **-** | **-** | Pass |
| NP  (No Рass) | **-** | **-** | NoPass |
| W  (Withdrawal) | - | - | Withdrawal |
| AW (Academic Withdrawal) |  |  | Academic Withdrawal |
| AU  (Audit) | - | - | Audit |
| Атт. |  | 30-60, 50-100 |  |
| Haven’t. |  | 0-29, 0-49 |  |
| R (Retake) | - | - | Retake |

*Discussed at a meeting of the Department of* molecular biology and genetics division *Protocol No \_\_ "\_\_" \_\_\_\_\_\_\_\_\_\_\_*

**Chief of the** molecular biology

and genetics division**, Professor Z.G. Aitasheva**

**Lecturer: A.B. Bigaliyev**

**Exams question**

1 Subject, tasks and methods of genetics. Periods of genetics.   
2. Hybridization method for the study of heredity.   
3 Monohybrid crossing. I, II Mendel's laws, their cytological basis.   
4 Di and poligibrids crossing. 3th Mendel's law and its cytological study. The general formula for splitting for independent inheritance of traits.   
5 Condition of Mendelian traits. Mendelian traits in man (autosomal dominant, autosomal recessive, examples).   
6 Types of interaction of alleles: dominance, overdominance, and codominance of multiple allelism phenomenon.   
7 Incomplete dominance. The definitions. Examples (see the nature of inheritance cystinuria, sickle-cell anemia, thalassemia, akatalazee).   
8 Multiple allelism (determining the cause of the process of evolution, the nature of the inside interaction between alleles). Examples.   
9 Inheritance of blood groups in AB0 systems.   
10 Inheritance and Rh factor (the system of Wiener and Fisher-Flight).

11 Medical value: incompatibility of peoples on blood groups and Rh factor.   
12 The complementary interaction of genes. The definitions. Nature of the splitting (9: 7) to consider specific examples.   
13 Epistasis. The definitions. Dominant and recessive epistasis. Know the concept of "epistatic gene" (suppressor gene, a gene-inhibitor) and "hypostatic gene."   
14 Dominant epistasis. Nature of the splitting (13: 3) to consider a concrete example.   
15 Recessive epistasis. The definitions. Consider the example of the Bombay phenomenon.   
16 Polymeric gene interactions. The definitions, examples. The additive effect of genes. Consider the examples: the inheritance of growth and color.   
17 Characteristics of Drosophila as a genetic object.   
18 Methods for chromosome mapping.   
19 The phenomenon of gens linkage. Linkage groups and their number. Crossingover. Probability of crossing over. Chromosome theory of heredity.

20, Inheritance of sexually linked:   
• list the loci of full and partial engagement with the X-chromosome;   
• golandric properties and the nature of their inheritance;

21 Properties limited and controlled the sexual. The definition, examples.   
22 Man as a specific object of genetic analysis.   
23 Genealogical research method. Rules for compiling genealogies cart. Analysis of pedigrees with monogenic inheritance of the character:   
• golandric type of inheritance;   
• properties of dominant and recessive type of inheritance;   
• know the characteristics of autosomal and X-linked inheritance type for both the dominant and recessive traits to;

24 Medical genetic counseling (problem, the indications for treatment, the stages of counseling).   
25 Cytogenetic method study of heredity. Karyotyping method.   
26 Cytological methods of rapid diagnosis:   
• methods for determining the X-sex chromatin (Barr bodies and "drumsticks");   
• methods for the determination of U-sex chromatin;   
27 The concepts of the methods of laboratory diagnosis of metabolic diseases (for example, phenylketonuria).   
28 Twins method of research. Concordance and discordance twins. Holzinger formula and its application. The role of genetic and environmental factors in the development of symptoms.

29 Population-statistical method for the study. The definition, stages of the study. Law Hardy - Weinberg and his position. Conditions of the law. Practical application of the law in human genetics.   
30 Dermatoglyphic method of research (definition, history of development, application). Formation of skin patterns in humans and their characterization.   
31 Dermatoglyphics with hereditary pathology examples.

32 Methods of prenatal diagnosis. Ultrasound and amniocentesis. The essence of the method and meaning.

33 Variability. Forms of variability (modification, combinative, genotypic). Determining the characteristic value in the evolution and ontogenesis.

34 Modification variability. Norm reaction, examples. The adaptive nature of the modifications, phenocopies.   
35 Mutational variability. Classification of mutations:   
• spontaneous and induced;   
• generative and somatic;   
• genomic, chromosomal aberrations and gene mutations;   
• lethal, half-lethal, neutral, positive;   
36 The mechanisms of spontaneous and induced mutagenesis. "Hot spots" mutations.   
37 Genomic mutations and their classification (poly-and geteroploidiya cells). Mechanism disorders.   
38 Geteroploidiya in the autosomes. Syndromes and methods for their diagnosis.   
39 Geteroploidii in the sex chromosomes. Syndromes and methods for their diagnosis.   
40 Chromosomal aberrations and their classification.   
41 Syndromes caused by disturbance of chromosome structure, and methods of diagnosis.   
42 Gene mutations and their classification.   
43 Molecular disease. The definitions, their classification:   
• monogenic diseases, types of inheritance, examples;   
• polygenic and multifactorial disease, examples;   
44 Metabolic diseases (fermentopatology). The definitions, types of inheritance examples.   
45 Phenylketonuria. Type inheritance mechanism of the disease, methods of diagnosis.   
46 Principles of treatment of hereditary diseases. Gene therapy.   
47 Mutagenic factors and their effects on the genetic apparatus of the cell. Komutagenic effects. The concept of anti-mutagenic, and reparogenasis desmutagens.   
48 Mechanisms for repairing damaged DNA structure: prereplicative repair (photoreactivation and excision repair).   
49 Replicative and post-replicative DNA repair.   
50 Stem cells and their characteristics.   
51 Modern methods of studying DNA. Sequencing.   
52 Diseases of the unconventional mode of inheritance: a mitochondrial disease.   
53 Famous scientists and their achievements: Schwann, Schleiden, Mendel, Correns, Chermak, de Fries, Morgan, Jacob Mono, Creek, Watson, Vavilov, Kolzov, Timofeev Resovskii.