



**APPROVED
EMD decision**

" 12 " 10 2021

Protocol No. 5

Chairman of the EMC, Vice-Rector,
candidate of pedagogical sciences,
associate professor Apeyova D.U.



SYLLABUS by discipline

CC 3.8.10. MEDICAL GENETICS

For students of the educational program, higher professional education in the specialty 560001 "General Medicine" (5-year education) in the specialty "Doctor"

Type of study work	Total hours
course	3
Semester	6
Number of weeks	18
Credits	2
The total complexity of the discipline	60
Classroom/practical studies (PS)	36
Student Independent Work (SIW)	24
Forms of control	
current control	Testing, oral questioning, written test
Frontier control	Testing
Midterm	Testing
Final control	exam
Semester rating by discipline:	Point-rating system

Information about the teacher of the academic discipline

Full Name	Otogonova Jyldyz Kadyrovna
Post	teacher
Academic degree	Candidate of medical sciences
Academic title	
Email address	
Location of the department (address)	KR, Bishkek, st. Shabdan Baatyr 128, floor 2
Telephone	0550030xxx
Consultation hours	11.00-13.30

Characteristics of the academic discipline

The purpose of studying the discipline The purpose of mastering the discipline "Medical Genetics" is to get students systematic theoretical and applied knowledge about the fundamental and applied aspects of medical genetics, to gain knowledge about the structure of the human genome and hereditary pathology caused by a violation of this structure, modern approaches to the diagnosis, prevention and therapy of hereditary diseases, to form a system of knowledge about the historical aspects of the formation of medical genetics in our country and in the world, directions and prospects of development; to form a system of knowledge about the mechanisms of formation and transmission of hereditary and congenital human pathology in a number of generations; to form a system of knowledge about the basics of human population

genetics; to form a system of knowledge about the clinical and genetic characteristics of frequent hereditary pathology and multifactorial diseases, methods of their diagnosis, approaches to prevention and therapy; to form knowledge about the methods used in medical genetics, acquisition of skills for solving genetic problems; to form readiness and ability to apply methods and technologies of analysis of genes and genome used in medical genetics.

Prerequisites of the discipline:

- Normal anatomy
- Normal physiology
- Basic pharmacology

Post-requirements of the discipline:

- Occupational diseases
- Fundamentals of clinical examinations in internal diseases
- Public health and healthcare
- Epidemiology

Learning outcomes of the discipline according to the RO GPP

The study of the discipline microbiology, virology and immunology will contribute to the achievement of learning outcomes (RE) GEP:

RE-3: analyze various causes (genetic, intrauterine, metabolic, toxic, microbiological, autoimmune, neoplastic, degenerative and traumatic) of painful and borderline conditions in the body.

Within the framework of this discipline, it is expected to achieve the following results of teaching the discipline, which are implemented within the framework of achieving competencies:

PC-14 -is capable and ready to make a diagnosis based on the results of biochemical and clinical studies, taking into account the course of pathology in organs, systems and in general;

Content of the discipline

№№	Name of topics
1.	Topic 1. Medical genetics: basic concepts, tasks, directions and prospects of development.
2.	Medical genetics: basic concepts, tasks, directions and prospects of development.
3.	Classical research methods in medical genetics: opportunities and limitations
4.	The current milestone (modular) control on topic 1.
5.	Topic 2. Molecular genetic foundations of human pathology
6.	The structure of the human genome: prerequisites for the formation of disorders leading to the development of hereditary diseases.
7.	Violations of matrix processes and regulation of genome expression as a cause of hereditary pathology.
8.	Modern classification and nomenclature of mutations. Mutation databases. The molecular basis of the pathogenicity of mutations and their implementation into pathology on the examples of frequent monogenic pathology with different types of inheritance
9.	Molecular and genetic foundations of cell division. Cytogenetic and molecular genetic mechanisms of the occurrence of chromosomal mutations. Molecular and genetic foundations of determination, differentiation and apoptosis
10.	Current milestone (modular) control on topic 2
11.	Topic 3. Hereditary and multifactorial human pathology: clinical, molecular and genetic characteristics.
12.	Modern approaches to the classification of congenital and hereditary pathology. Clinical polymorphism and genetic heterogeneity of human pathology
13.	Congenital malformations: mechanisms of formation of malformations. Chromosomal pathology.
14.	Violations of the formation of sex.
15.	Clinical and genetic characteristics of hereditary diseases of the nervous system. Criteria and methods of diagnosis
16.	Clinical and genetic characteristics and basic approaches to the classification of hereditary metabolic diseases

17.	Splicing disorders as a molecular mechanism of pathology. Bioinformatics of RNA splicing: variants of splicing, assessment of norm and pathology. Splicing as a tool for determining exons.
18.	Polymerase chain reaction: physico-chemical mechanisms. Stages and components of PCR. The main types and modifications of PCR (real-time PCR, reverse transcript PCR, multiplex PCR, methyl-sensitive and methyl-specific PCR, adapter -mediated PCR, MLPA): physico-chemical characteristics, stages and components, capabilities and limitations
19.	Current milestone (modular) control on topic 3
20.	Topic 4. Modern problems of diagnosis, treatment and prevention of hereditary pathology.
21.	Genomic technologies in medical genetics: methods of gene analysis
22.	Analysis and interpretation of the results of molecular genetic studies
23.	Software for the design of oligonucleotide primers and design quality control. Criteria for the quality of primer design. Design of oligonucleotide primers for PCR: for the search for mutations by Sanger sequencing methods, for the analysis of restriction fragment length polymorphism (PDRF), with the creation of an artificial restriction recognition site
24.	Genomic technologies in medical genetics: methods of genome analysis.
25.	DNA sequencing by Sanger: automatic genetic analyzer, analysis of the results using the "Chromas" software. Analysis of Sanger DNA sequencing results using the UCSC genomic browser
26.	High-performance parallel DNA sequencing as an effective method of diagnosing molecular and cellular disorders in hereditary pathology. Principles of analyzing the results of high-performance parallel DNA sequencing: abstract of genetic variants, genomic browser 3 T + + + 12 Integrative Genomics Viewer (IGV
27.	Design of oligonucleotide primers for multipath PCR, for multipath methyl-sensitive PCR.
28.	Software for the design of TaqMan probes and "scorpion" primers. Design of "scorpion" primers for real-time PCR
29.	Methods of prevention and therapy of hereditary pathology (including Prenatal diagnosis and screening: methods, indications, development prospects. Modern approaches to the detection of molecular and cellular disorders leading to chromosomal pathology and pathology of pregnancy. Preimplantation genetic diagnosis (PGD).
30.	DNA fragment analysis: interpretation of the results. Comparative characteristics of the interpretation of the results of DNA sequencing by Sanger and DNA fragment analysis
31.	Methods of prevention and therapy of hereditary pathology (including neonatal screening; approaches to the development of pathogenetic therapy on the example of NBO)
32.	Current milestone (modular) control on topic 4
33.	Current final control for all sections

List of main and additional literature:

Main literature:

1. Cell Biology and Genetics M. Stubbs, N. Suleyman. - 4th ed. - Edinburgh etc. Mosby Elsevier, 2015
2. Clinical genetics: [study. for higher Prof. education] N. P. Bochkov, V. P. Puzyrev, S. A. Smirnikhina; edited by N. P. Bochkov. 4th ed., supplement and revision. – Moscow: GEOTAR-Media, 2013

Additional literature:

1. Clinical genetics: genomics and proteomics of hereditary pathology: [textbook. manual for universities] Mutovin, G. R. 3rd ed., reprint. and additional–M.: GEOTAR-Media, 2010
2. Hereditary diseases: national hands. [Alekseev L. P. et al.]; chief editor N. P. Bochkov [et al.]. Moscow: GEOTAR-Media, 2012
3. Genetics: [textbook for universities] V. I. Ivanova. N. V. Moscow: Akademkniga, 2007
4. Genes Lewin B. [Text] M.: BINOM. Laboratory of Knowledge, 2012.

Internet resources:

<http://www.edu.ru>
<http://www.medicina.ru>
<http://www.elibrary.ru>
<http://www.genenames.org>
[http //www.journals.uchicago.edu/JAD/home.html](http://www.journals.uchicago.edu/JAD/home.html)

Monitoring and evaluation of learning outcomes

The content of the rating system for assessing student performance

The rating assessment of students' knowledge in each academic discipline, regardless of its total labor intensity, is determined on a 100 (one hundred) - point scale and includes current, boundary, intermediate and final control.

The distribution of rating scores between types of control is established in the following ratio (according to the table of the score-rating system of assessments):

Form of control				
current (CC)*	boundary control (BC)**	mid-term exams (MC)***	Final /exam (FE)	Discipline Rating (RD)
0-100 points	0-100 points	0-100 points	0-100 points	0-100 points, with the translation of points into a letter designation

Note:

* $TK(middle) = \frac{\sum_1^n \times point}{\sum_1^n}$, where n is the number of types of classroom and extracurricular work of students in the discipline;

** $PK(middle) = \frac{\sum_1^n credit \times point}{\sum_1^n credits}$, where n is the number of modules (credits) in the discipline;

*** $ПК(middle) = \frac{\sum_1^n \times point}{\sum_1^n}$, where n is the number of intermediate controls (2 controls per semester: in the middle and at the end of the semester) by discipline;

****ИК – examination conducted at the end of the study of the discipline

;

*****РД = $\frac{TK_{ср} + PK_{ср} + ПК_{ср} + ИК}{4}$, the final rating of the results of all types of control at the end of the discipline;

GPA = $\frac{\sum_1^n \times балл}{\sum_1^n}$ where, n is the number of disciplines in the semester (for the past period of study).

A student who has not passed the current, boundary and intermediate controls to the final control (exam) is not allowed.

The current control is carried out during the period of classroom and independent work of the student on time according to the schedule, at the end of the study of the discipline, the average score of the current control (CC) is calculated. *Forms of current control can be:*

- testing (written or computerized);
- performance of individual homework assignments, abstracts and essays;
- student's work in practical (seminar) classes;
- various types of colloquia (oral, written, combined, express, etc.);
- control of performance and verification of reporting on laboratory work;
- visiting lectures and practical (seminar, laboratory) classes;
- Incentive rating (up to 10 points).

Other forms of current monitoring of results are also possible, which are determined by the teachers of the department and recorded in the work program of the discipline.

The frontier control is carried out in order to determine the results of the student's development of one credit (module) as a whole. *Frontier control* should be carried out only in writing, at the end of the study of the discipline, the average score of boundary control (BC) is calculated. As forms of *frontier control* of the training module, you can use:

- testing (including computer testing);
- interview with written fixation of students' answers;
- test.

Other forms of intermediate control of results are also possible.

Intermediate control (mid-term exams) is carried out in order to check the completeness of knowledge and skills in the material in the middle and end of the semester (2 times per semester) of studying the discipline, by the end of the study of the discipline, the average score of intermediate control (PCsr) is calculated, *forms of intermediate control (mid-term exams) can be:*

- testing (including computer testing);
- interview with written fixation of students' answers;
- test.

Other forms of intermediate control of results are also possible.

The final control is carried out during the session, by conducting an exam, it can be carried out in the following forms:

- testing (including computer testing);
- written exam (ticketing system).

Correspondence of the point-rating system of assessments used by the institute and the assessments of the European system for the transfer of credit units, labor intensity (ECTS)

Grade						Criterion
System of letters	digital system	Traditional system	Points (%)	Scored points (max - 100)	Evaluation by discipline without an exam	
A	4	5	95-100	95-100	Credited/ passed	"Excellent" - deserves a student who has shown a deep, systematic and comprehensive knowledge of the educational material, who freely performs practical tasks, who has mastered the recommended basic and additional literature on the discipline
A-	3,67		90-94	90-94		"Excellent" - deserves a student who has shown a deep, systematic and comprehensive knowledge of the educational material, who freely performs practical tasks, who has mastered the recommended basic literature on the discipline, but is not familiar with additional literature
B+	3,33	4	85-89	70-89		"Good" - exhibited to a student who has shown a systematic and comprehensive knowledge of the educational material, able to independently replenish and update this knowledge in the course of training, performing practical tasks, familiar with the main literature on the discipline
B	3,0		80-84			"Good" is given to a student who has shown a systematic and comprehensive knowledge of the educational material, who is able to independently replenish this knowledge in the course of training, performing practical tasks, but not fully familiar with the main literature on the discipline
B-	2,67		75-79			"Good" - is given to a student who has shown the systematic nature of knowledge in the discipline, who is able to independently replenish this knowledge in the course of training, performing practical tasks, but not fully familiar with the main literature on the discipline
C+	2,33		70-74			"Satisfactory" - is given to a student who does not have a systematic nature of knowledge in the discipline, who is not capable of independently replenishing and updating knowledge in the course of further education, performing practical tasks with errors
C	2,0	3	65-69	50-69		"Satisfactory" - is given to a student who made mistakes in completing assignments, but who has the necessary knowledge to eliminate them under the guidance of a teacher
C-	1,67		60-64			"Satisfactory" - is set to a student who made errors in the performance of tasks, but who has the possible knowledge to eliminate them under the guidance of a teacher
D+	1,33		55-59			"Satisfactory" - is set to a student who made errors in the performance of tasks, who does not have the necessary knowledge to eliminate them
D-	1,0		50-54			"Satisfactory" - is given to a student who has made significant errors in the performance of tasks, who does not have the necessary knowledge to eliminate them
FX	0,5	2	25-49	Less of 50	not credited/not passed	"Unsatisfactory" - is set to a student who has not completed the task, does not have the necessary knowledge to eliminate them
F	0		0-24			"Unsatisfactory" - is set to a student who has not completed the task, does not have the necessary knowledge to eliminate them, even under the guidance of a teacher

Academic achievement requirements:

Attendance by students of all classroom classes without delay is mandatory.

In case of absence, classes are worked out in the order established by the dean's office.

If there are three passes, the teacher has the right not to allow the student to attend classes until the issue is administratively resolved.

If the absence of classes is more than 20.0% of the total number of classes, the student automatically enters the summer semester.

Note to the student:

- ✓ regularly review lecture material;
- ✓ Do not be late and do not miss classes;
- ✓ work off missed classes if you have permission from the dean's office;
- ✓ Actively participate in the classroom (individually and in groups;)
- ✓ timely and fully complete homework assignments;
- ✓ submit all assignments within the time specified by the teacher;
- ✓ independently study the material in the library and at home;
- ✓ timely and accurately fulfill the tasks of the teacher, individual tasks for the IWS to achieve learning outcomes;
- ✓ to master the basic and additional literature necessary for the study of the discipline;
- ✓ performing tasks, the student should not copy or reproduce the work of other students, scientists, practitioners, plagiarism;
- ✓ develop their intellectual and oratory skills;

In case of non-compliance with the requirements of the Memo, the student will be penalized in the form of deducting points (one point for each violated item).

If the requirements of the Memo are fully met, the student is encouraged in the form of an additional 10 points to the final control in the discipline.

Academic Integrity, Conduct and Ethics Policy:

- turn off your cell phone during class;
- Be polite;
- respect other people's opinions;
- formulate objections in the correct form;
- do not shout or raise your voice in the audience;
- independently complete all semester assignments;
- Eliminate plagiarism from your practice;

Methodical instructions.

It is recommended to organize the time required to study the discipline as follows:

When preparing for a practical lesson, you must first read the abstract with the teacher's explanations.

When performing exercises, you must first understand what you want to do in the exercise, then proceed to its implementation.

Literature work. The theoretical material of the course becomes more understandable when books are studied in addition to the abstract. After studying the main topic, it is recommended to perform several exercises.

Preparation for boundary and intermediate controls. In preparation for the boundary and intermediate control, it is necessary to study the theory: the definitions of all concepts before understanding the material and independently do several exercises.

Independent work of students is organized on all studied topics of each section. Independent work is carried out in the form of:

- work in Internet sites;
- work with basic and additional literature;
- fulfillment of written assignments;
- preparation of reports, abstracts, tables and posters on