AL-FARABI KAZAKH NATIONAL UNIVERSITY
Faculty of Medicine and Healthcare
Higher School of Medicine
Department of Fundamental Medicine

PROGRAM
of final exam on discipline
Molecular bases of pathology
(medical genetics, pharmacology)"
(10 credits)
2024-2025 academic year

Expected outcomes: Students in the final exam must demonstrate the ability to:
1. Apply knowledge about molecular and genetic aspects of genetically determined diseases (chromosomal, monogenic, polygenic); understand the principles of genetic diagnostics and medical genetic counseling
2. Apply knowledge of molecular-genetic, biochemical mechanisms of the body's response to drugs and biologically active compounds
3. Understand the biochemical processes in the main pathological conditions and genetically determined diseases
4. Interpret the results of specific molecular genetic diagnostic methods
5. Understand the role of relevant risk factors of diseases for decision-making with a view to their prevention
6. Integrate knowledge on human genetics, immune response, biochemical processes and the interaction of micro and macro-organism for the purposes of diagnosis and personalized treatment of human pathology
7. Know the pharmacokinetic parameters, mechanisms of absorption and biotransformation of drugs
8. Apply knowledge of pharmacodynamics and mechanisms of action of drugs in the main pathological processes (affecting the acid-base state, hemostasis and hematopoiesis, inflammation, infectious process, allergies, autoimmunity, onco-process). Know the types of undesirable side reactions and understand the possibilities of their correction
9. Demonstrate the ability to identify learning gaps and create strategies to enhance one’s own knowledge and skills
10. Effectively communicate with other students and teachers regarding medical and scientific information, articulate their opinions clearly when discussing and work effectively as a member of the team

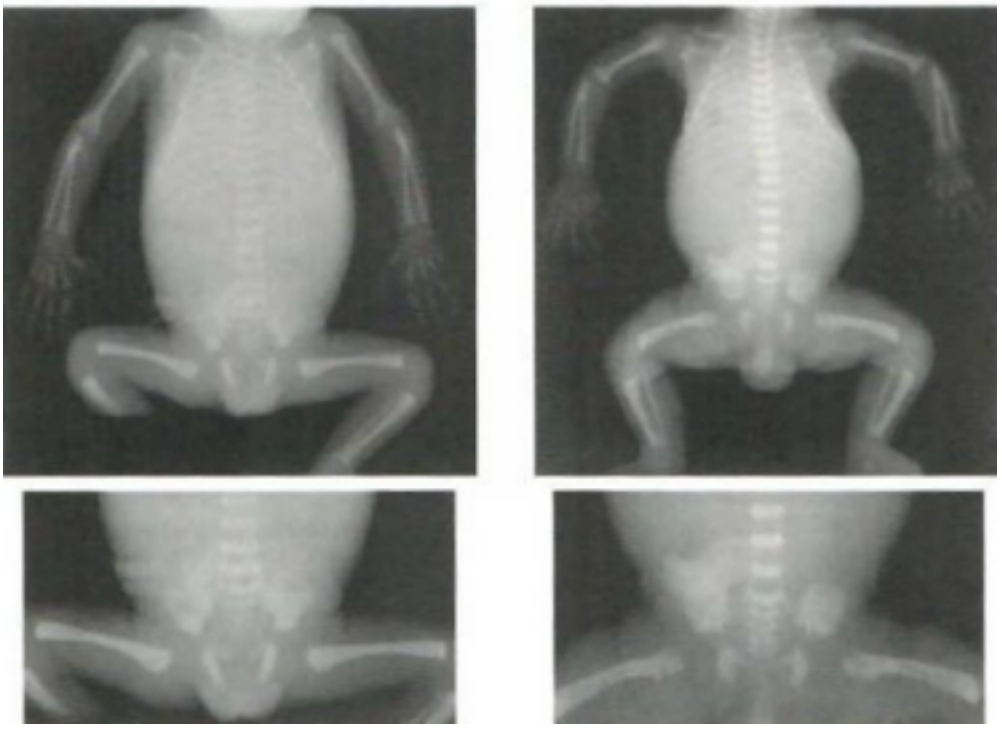
I MEDICAL GENETICS
(3 ECTS)

Approved final exam form - written exam
Topics included in final exam:
1. Introduction to Medical Genetics. Chromosomal disorders (Down syndrome, Patau syndromes, Edwards syndrome, Cri du chat syndrome)
2. Sex Chromosome disorders (Trisomy-X, Y-disomy, Klinefelter syndrome, and Turner syndrome). Summary of chromosomal diseases
3. Mendelian classic disorders: autosomal inheritance (Phenylketonuria, galactosemia, alkaptonuria, fructosuria, Cystic fibrosis, Marfan syndrome, Achondroplasia, Wilson-Konovalov syndrome, Hypertrichosis)
4. Mendelian classic disorders: sex-linked inheritance (hemophilia, color blindness, Ichthyosis, Lesch–Nyhan syndrome, Duchenne Muscular Dystrophy, Vitamin-resistant rickets)

5. Biochemical bases of hereditary metabolic disorders
6. Biochemical disorders in lipid metabolism enzymopathy
7. Non-mendelian genetic disorders (Prader-Willi syndrome, Angelman syndrome, Huntington's disease)
8. Fundamentals of population genetics
9. Pharmacogenetics
10. Polygenic multifactorial disorders. (Diabetes mellitus, Schizophrenia, Family Hypercholesterolemia, Arterial hypertension)
11. Cancer Genetics and Genomics (breast cancer)
12. Metabolic aspects of cancer
13. Polygenic disorders: developmental malformation (Neural tube defect, Cleft lip and cleft palate, hip dysplasia).
14. Genetic counseling. Genetic testing, prevention and treatment.

After completing this course students will be able to:
1. apply knowledge about molecular and genetic aspects of genetically determined diseases (chromosomal, monogenic, polygenic); understand the principles of genetic diagnostics and medical genetic counseling.
2. apply knowledge of molecular-genetic, biochemical mechanisms of the body's response to drugs and biologically active compounds.
3. understand the biochemical processes in the main pathological conditions and genetically determined diseases.

Sample typology of exam assignments
Case



P.S., a 30-year-old healthy woman, was 27 weeks pregnant with her first child. A fetal ultrasound examination at 26 weeks' gestation identified a female fetus with macrocephaly and rhizomelia (shortening of proximal segments of extremities). P.S.'s spouse was 45 years of age and healthy; he had three healthy children from a previous relationship. Neither parent has a family history of skeletal dysplasia, birth defects, or genetic disorders. The obstetrician explained to the parents that their fetus had the features of achondroplasia. The infant girl was delivered at 38 weeks' gestation by cesarean section. She had the physical and radiographic features of achondroplasia, including

frontal bossing, megalencephaly, midface hypoplasia, lumbar kyphosis, limited elbow extension, rhizomelia, trident hands, brachydactyly, and hypotonia. Consistent with her physical features, DNA testing identified an 1138G>A mutation leading to a glycine to arginine substitution at codon 380 (Giy380Arg) in the fibroblast growth factor receptor 3 gene (FGFR:JJ.)

Types of Questions:

1. Explain main genetic data/symbols given for this disease in OMIM (I level)
2. What are the main features of inheritance patterns of this genetic group of diseases? (I level)
3. Write about the main causes leading to this disease? (I level)
4. What is the epidemiology of this disease? Prevalence rate? (I level)
5. What genetic variants of this disease do you know? (I level)
6. What phenotypic forms of this disease are known? Explain causes of phenotype diversity. (I level)
7. Describe (sketch) the proband family tree? (II level)
8. Explain the probably mechanism of mutations in this case /this disease in general / this type of diseases… (II level)
9. Explain how phenotypic severity of this disease in general / this type of diseases… correlates with the type of mutation (II level)
10. Discuss probable religious, cultural, social and ethical beliefs and understanding might affect the decisions the patient/ family would make (II level)
11. Discuss probable ethical and legal issues regarding diagnosis /prevention / treatment for this case /this disease in general/this type of diseases...(II level)
12. Explain clinical polymorphism regarding types of mutation of this disease (II level)
13. What is the correlation between phenotype and genotype? (II level)
14. Propose diagnosis. Explain which group of genetic diseases belongs this case to? (III level)
15. Propose and explain risk assessment strategy for this family/proband/siblings...(III level)
16. Propose genetic mechanisms that affect the proband phenotype and explain your thoughts? (III level)
17. Propose methods of diagnosis /prevention / treatment for this case /this disease in general/this type of diseases and explain your strategy (III level)
18. Explain results of genetic testing (III level)
19. Name other disorders that are characterized by prevalence rate like this. What types of mutations are associated with these disorders? (III level)
20. Compare and contrast the pathological mechanisms of this disease in general/this type of diseases and correlate them with clinical presentation (III level)

II. GENERAL PHARMACOLOGY

(4 ECTS)

Approved final exam form - written exam

Topics included in final exam on General Pharmacology

1. Introduction to Pharmacology
2. Pharmacokinetics
3. Pharmacodynamics
4. PNS. Cholinergic drugs
5. Cholinoblockers
6. PNS. Adrenergic drugs
7. Adrenoblockers
8. Hypnotics
9. Anti allergics SAIDS
10. Antihypertensive drugs. Antianginal drugs

11. Pharmacology of hematopoiesis and hemostasis system
12. Pharmacology of ES. Pancreas, diabetes.
13. Anti Inflammatory drugs. NSAIDs, SAIDs
14. Analgesics
15. Antibiotics. Classification. Beta-lactams. Macrolides. Tetracyclines, Aminoglycosides. Peptide antibiotics
16. Antibiotics. Nitroimidazoles and nitrofurans. Quinolones. Sulfonamides.TB
17. Antiviral drugs. Treatment of HIV infection. Antifungal preparations.

List of drugs to repeat before final examination

1. Epinephrine
2. Norepinephrine
3. Enalapril
4. Acetylsalicylic acid
5. Metoprolol
6. Losartan
7. Hydrochlorothiazide
8. Insulin Actrapid
9. Insulin NPH
10. Fluticasone
11. Salbutamol (Albuterol)
12. Amoxicillin/clavulanate
13. Cortisol (hydrocortisone)
14. Spironolactone
15. Metformin
16. Neostigmine
17. Dexamethasone
18. Heparin
19. Warfarin
20. Furosemide
21. Ceftriaxone
22. Prazosine
23. Nifedipine
24. Isoniazid
25. Streptomycin

Metronidazole
Ciprofloxacin
28. Azithromycin
29. Gliclazide
30. Fluconazole
31. Acyclovir
32. Amphotericin B
33. Rifampicin
34. Furazolidone
35. Co-trimoxazole
36. Pioglitazone
37. Morphine
38. Diclofenac
39. Rivaroxaban
40. Nitroglycerin
41. Doxycycline
42. Vancomycin
43 Interferon
44. Lidocaine
45. Phenobarbital
46. Zolpidem
47. Loratadine
48. Fluticasone
49. Nedocromil sodium
50. Suprastin

After completing this course students will be able to:

1. know the pharmacokinetic parameters, mechanisms of absorption and biotransformation of drugs.
2. apply knowledge of pharmacodynamics and mechanisms of action of drugs in the main pathological processes (affecting the hemostasis and hematopoiesis, inflammation, infectious process, hypertension, hyperglycemia, hypotension). Know the types of undesirable side reactions and understand the possibilities of their correction.
3. demonstrate the ability to identify learning gaps and create strategies to enhance one's own knowledge and skills.

Approximate typology of exam tasks

Part1.

Case

A 75-year-old patient, whose elevated blood pressure was successfully controlled by taking loop diuretic, recently began complaining of muscle weakness, paresthesia, dyspeptic disorders,

tachycardia. Because of these side effects, he cannot cope with his daily work.

Types of questions:
1. Tell the mechanism of action of the drug
2. Explain the mechanism of drug side effects
3. Explain the mechanism of drug interaction
4. Explain the pharmacologic basis of switching one drug to another
5. Compare mechanism of action of two drugs with similar effect
6. Propose agonist/antagonist of hormone
7. Explain the mechanism of action of hormone, and how drugs can affect it 8. Describe the drug – partial agonist, agonist, inverse agonist
9. List the additional medication, that can help in this condition
10. Explain how the side effects are linked with drug mechanism of action
11. Call the name of the medicine

Part 2.
Write a Prescription for the following drugs:
1. Enalapril
2. Amoxicillin/clavulanate
3. Epinephrine

Guidelines for exam conducted offline in the classroom.
WRITTEN EXAM:
TRADITIONAL - ANSWERS TO QUESTIONS.

The process of taking a written exam by the student involves the automatic creation of an exam ticket for the student, to which you must form a written answer by direct handwriting.

Exam Technology Instructions

TRADITIONAL - ANSWERS TO QUESTIONS.

The process of taking a written exam by the student involves the automatic creation of an exam ticket for the student, to which you must form a written answer by direct handwriting.

Exam Technology Instructions

1. The duration of the exam is exactly 2 hours.
2. Written exams are administered according to the approved schedule.
3. Students may enter the auditorium where the written exam is administered only with an ID card (Passport or student ID card). The presence of persons not participating in the examination procedure is prohibited.
4. The proctor reconciles the identification document with the admission permission slip. A student who has a discipline clearance rating of less than 50% is not allowed to take the written exam.
5. The proctor (calls the names from the list and sits them down according to the list) starts them in the auditorium.
6. Late students are not allowed to take the exam.
7. Proctor gives each student an answer sheet (if necessary, the student may take an additional answer sheet) and gives the student the opportunity to choose a ticket for the discipline being passed (the text of the ticket should not be visible to the student).

8. Students present at the exam must sign the admission form.
9. The start and end times of the written exam are recorded on the blackboard.
10. During the written exam, students' questions on the content of the examination tickets are not considered.
11. If the student does not comply with the established requirements at the exam: uses crib notes, mobile and other devices, allows disciplinary violations, disturbs other students with their actions, the proctor has the right to remove student from the audience. In this case, an act of violation of the examination procedure is drawn up, the answer sheet is annulled by crossing out diagonally, the mark "Removed for violation" is made in the admission sheet, "0" points will be given in the sheet.

12. it is allowed for a student to visit the restroom no more than 1 time per hour, lasting no more than 5 minutes. If frequent visits to the restroom are required (for example, due to health conditions), the student must undergo a medical examination, and the exam is counted as the student's absence from the exam.
13. At the end of the exam, the student must turn in his/her ticket and answer sheet.

Scale of response quality

Evaluation	**Criteria**	**Scale, points**
Excellent	1. All key aspects included and presented logically; 2. High accuracy (relevance, without redundancy) and consistent focus on question; 3. Excellent integration of theoretical issues; 4. Provision of relevant examples; 5. In-depth analysis and theoretical justification of given problem (if applicable), all key aspects identified and interpreted; 6. Fluency in use of professional terminology	90 - 100
Good	1. All key aspects included and presented logically; 2. Consistent focus on question with satisfactory accuracy, and relevance, and/or some redundancy; 3. Satisfactory integration of theoretical issues; 4. Lack of examples; 5. Satisfactory analysis and theoretical justification of given problem (if applicable), most key aspects identified and interpreted; 6. Correct use of professional terminology	70 - 89
Satisfactory	1. Most key aspects included; 2. Satisfactory focus on question - some lapses of relevance and/or noticeably redundancy; 3. Theoretical issues presented without noticeably integration; 4. Provision of unsuccessful examples or no examples; 5. Some analysis and theoretical justification of given problem (if	50 - 69

	applicable), most key aspects identified and interpreted; 6. Correct use of professional terminology	
Unsatisfactory (FX)	1. Most key aspects missed; 2. Lack of focus on question - no relevance and notable redundancy; 3. Some theoretical issues presented in someway; 4. No or irrelevant examples; 5. Some analysis and theoretical justification of a given problem (if applicable), most key aspects missed; 6. Lapses in use of professional terminology	25 - 49
Failed	1. Most or all key aspects missed; 2. No focus on question, irrelevant information; 3. Theoretical issues missed or superficial; 4. No or irrelevant examples; 5. No analysis and no theoretical justification of a given problem (if applicable), most key aspects missed; 6. Lapses in use of professional terminology	0-24

Grading system

Letter Grade	**The digital equivalent of points**	**% content**	**Traditional system assessment**
A	4,0	95-100	Excellent
A-	3,67	90-94	
B+	3,33	85-89	Good
B	3,0	80-84	
B-	2,67	75-79	
C+	2,33	70-74	
C	2,0	65-69	Satisfactory
C-	1,67	60-64	
D+	1,33	55-59	
D-	1,0	50-54	
FX	0	25-49	Unsatisfactory
F	0	0-24	Unsatisfactory
I (Incomplete)	0	-	“Discipline is not completed” *(not taken into account when*

			calculating GPA)

Basic literature:

1. Thompson & Thompson genetics in medicine (2016) Robert L. Nussbaum, Roderick R. McInnes, Huntington F. Willard, Ada Hamosh. Philadelphia, PA: Elsevier
2. Basic & Clinical Pharmacology [Electronic resource]: collection / ed.: B. G. Katzung, A. J. Trevor. - 13th ed. - New York; Chicago; San Francisco: McGraw-Hill Education, 2015. - 1837 p. - ISBN 978-0-07-182641-9: 0.00

Additional literature:

1. Jorde, L.B. et al. (2016) Medical Genetics. Philadelphia, PA: Elsevier
2. Emery's Elements of Medical Genetics (2017) Turnpenny, P.D., Ellard S. 15th Edition, Elsevier
3. Hartwell, L. et al (2017) Genetics: from genes to genomes, 6th edition. New York, NY: McGrawHill Education
4. USMLE Step 1 Lecture Notes (2017): Biochemistry and Medical Genetics. Kaplan Publishing

WWW resources:

1. OMIM® Online Mendelian Inheritance in Man® An Online Catalog of Human Genes and Genetic Disorders https://www.omim.org/
2. The Genetic Testing Registry (GTR®) https://www.ncbi.nlm.nih.gov/gtr/
3. Genetics Home Reference. https://ghr.nlm.nih.gov/resources
4. ClinGen: Clinical Genome Resource https://www.clinicalgenome.org/
5. Learn.Genetics https://learn.genetics.utah.edu/content/basics/
6. Clinical Genetic Education Resources (Courses and Lectures) https://www.kumc.edu/gec/prof/genecour.html
7. Genomics Education Program. https://www.genomicseducation.hee.nhs.uk
8. ELSEVIER "Clinical learning" training program, 2018
9. https://www.msdmanuals.com/professional/clinical-pharmacolog