

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

**PROGRAM**  
**of final exam on discipline**  
**MZiB2216 "Mechanisms of Defense and Disease (medical genetics, microbiology,**  
**pharmacology)"**  
**(10 credits)**  
**Autumn semester, 2022-2023 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. Apply knowledge of the infectious process and its features in various types of human pathogens, apply knowledge of immunodiagnostics of infectious diseases, apply knowledge of immunoprophylaxis, demonstrate an understanding of the principles of infection control and biosafety
5. Interpret the results of specific molecular genetic diagnostic methods
6. Understand the role of relevant risk factors of diseases for decision-making with a view to their prevention
7. 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
8. Know the pharmacokinetic parameters, mechanisms of absorption and biotransformation of drugs
9. 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
10. Demonstrate the ability to identify learning gaps and create strategies to enhance one's own knowledge and skills
11. 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) 7-8. Non-mendelian genetic disorders (Prader-Willi syndrome, Angelman syndrome, Huntington's disease)

5. Fundamentals of population genetics

6. Polygenic multifactorial disorders. (Diabetes mellitus, Schizophrenia, Family Hypercholesterolemia, Arterial hypertension)

7. Cancer Genetics and Genomics (breast cancer)

8. Polygenic disorders: developmental malformation (Neural tube defect, Cleft lip and cleft palate, hip dysplasia).

**After completing this course students will be able to:**

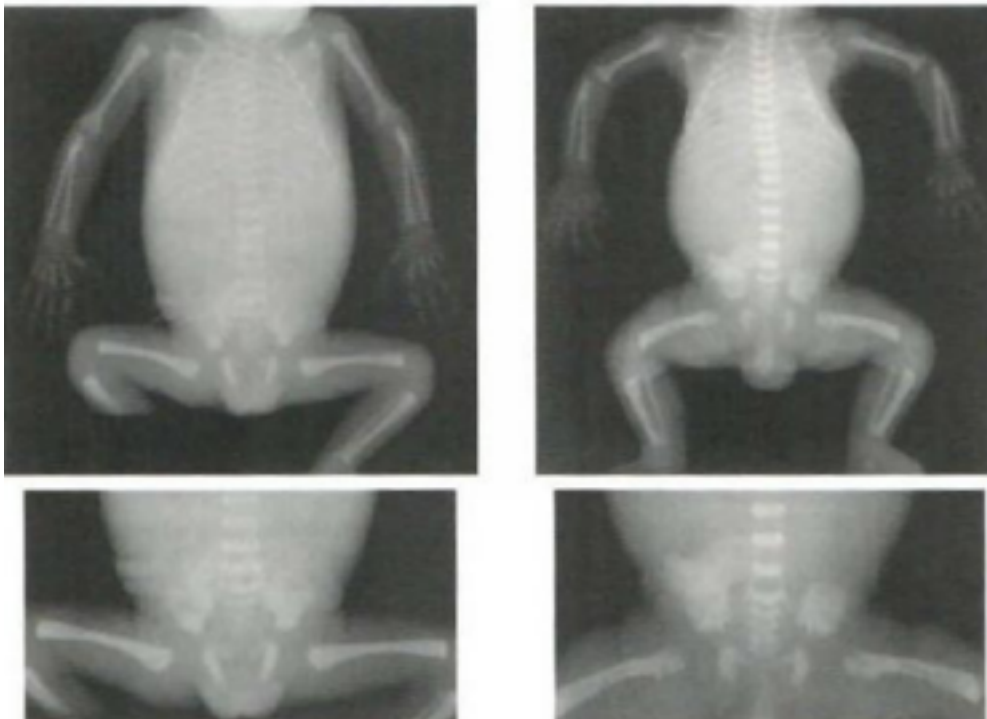
1. apply knowledge about molecular and genetic aspects of genetically determined diseases (chromosomal, monogenic, polygenic); understand the principles of genetic diagnostics and medical genetic counseling.

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 (Gly380Arg) in the fibroblast growth factor receptor 3 gene (FGFR:JJ.)

**Types of Questions. 5 of them to choose (2 from- of level I, 2- of level II, 1- of level 3):**

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

**II MEDICAL MICROBIOLOGY**  
(3 ECTS)

**Approved final exam form - written exam**

**Topics included in final exam:**

1. Gram-positive cocci. Microbiological diagnostics. Filling the staphylococcal infection research algorithm. The rules for the collection and delivery of material for infectious and somatic diseases caused by gram-positive cocci. Principles of treatment and prevention. Gram-negative cocci. Microbiological diagnostics. Filling the research algorithm for meningococcal infection. The rules for the collection and delivery of material for infectious and somatic diseases caused by gram-negative cocci. Principles of treatment and prevention
2. Isolation of a pure culture of enterobacteria (1-4 days of the study). Escherichia. Shigella. Vibrios. Diseases caused. Features of microbiological diagnosis in connection with the pathogenesis of diseases. Principles of treatment, prevention
3. Salmonella. Features of microbiological diagnosis in connection with the pathogenesis of caused diseases. Principles of treatment, prevention. Differential diagnosis of bacteria of the intestinal group. Campylobacter. Helicobacter. Features of microbiological diagnosis in connection with the pathogenesis of diseases. Principles of treatment, prevention
4. The causative agents of zoonotic infections. Brucellosis, plague, anthrax, tularemia. Features of microbiological diagnosis in connection with the pathogenesis of diseases. Statement of the reaction

- of Ascoli, Hedelson, Wright. Interpretation of the results. Principles of treatment, prevention
5. Pathogenic and conditionally pathogenic corynebacterium. Bordetella. Algorithm for laboratory diagnosis of diphtheria, pertussis and pertussis. Features of microbiological diagnosis in connection with the pathogenesis of diseases. Formulation of the Ouchterlony reaction. Interpretation of the results. Principles of treatment, prevention
  6. Pathogenic and opportunistic mycobacteria. Tuberculosis. Features of microbiological diagnosis in connection with the pathogenesis of diseases. Algorithm for laboratory diagnosis of tuberculosis. Principles of treatment, prevention  
Leprosy. Features of microbiological diagnosis in connection with the pathogenesis of diseases. Principles of treatment, prevention
  7. Pathogens of sexually transmitted diseases. Spirochetes. Mycoplasmas. Chlamydia Algorithm for laboratory diagnosis of sexually transmitted diseases. Features of microbiological diagnosis in connection with the pathogenesis of diseases. Principles of treatment, prevention
  8. The causative agents of anaerobic infections. Algorithm for laboratory diagnosis of anaerobic infections. Features of microbiological diagnosis in communication with the pathogenesis of diseases. Principles of treatment, prevention. Rickettsia, Borrelia. Features of microbiological diagnosis in connection with the pathogenesis of diseases. Principles of treatment, prevention
  9. Adenoviruses. Poxviruses. Rhabdoviruses. Role in human pathology. The principles of treatment. Prevention
  10. Orthomyxoviruses (influenza virus). Paramyxoviruses (viruses of parainfluenza, mumps, measles, respiratory syncytial virus). Statement of RGA, RTGA, RTGA in paired sera. Interpretation of the results
  11. Picornaviruses - causative agents of poliomyelitis, Coxsackie viruses, ECHO. Principles of treatment, prevention. Statement of reaction of color test. Interpretation of the results. Color sample mechanism Arboviruses. Role in human pathology. The principles of treatment. Prevention Rubella virus. Role in the pathology of pregnant women. Principles of treatment, prevention
  12. AIDS virus. ELISA for the diagnosis of HIV infection. Interpretation of the results. Principles of treatment, prevention. Oncoviruses. Principles of treatment, prevention. CMV infection. Role in human pathology. The principles of treatment. Prevention
  13. Hepatitis A, B, C. viruses. Treatment principles, prevention. Herpes viruses (alpha beta, gamma herpes viruses). Principles of treatment, prevention
  14. Fungal infections or mycoses. Candidiasis, Cryptococcosis, Aspergillosis, Blastomycosis. Laboratory diagnostic, treatment principles, prevention.
  15. Nosocomial diseases. Classification, risks, prevention, clinical cases.

### **Approximate typology of exam tasks**

**Every ticket has 3 questions.**

**Practical questions:**

#1 (max 40% or 12 points)

#2 (max 30% or 9 points)

**Theoretical question:**

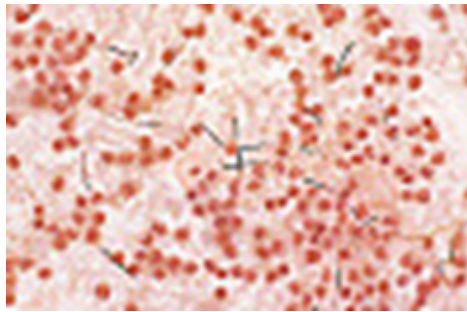
#3 (max 30% or 9 points)

Total mark: 100% or 30 points

#### **Case**

A suspicious envelope arrived for sorting at rural post office. The envelope was opened and found to contain white powder. Approximately two days later, the postal worker who handled the letter developed cutaneous boils, which were and 1 to 5 cm in diameter with central necrosis and eschars. He and his wife also developed a mild nonproductive cough with fatigue, myalgia for 72 hours, followed by severe dyspnea, diaphoresis and cyanosis. Temperature of 39.5°C, pulse 105/min, respiration 25/min, and blood pressure 85/45mm Hg. Crackles were heard at the lung bases. A

chest xray shows a widened mediastinum and small pleural effusions. WBC count of 13,130/mm<sup>3</sup>, hemoglobin 13.7g/dL, hematocrit 41.2%, MCV 91 um<sup>3</sup>, and platelet count 244,000/mm<sup>3</sup>. Both died despite antibiotic therapy. Several cattle, horses, and sheep on the postal worker's farm also died.



### **Types of Questions.**

- 1) Based on these clinical findings, what is the likely causative agent?
- 2) Describe the microbiological diagnostics (from samples until identification). Which specimen are appropriate samples for anaerobe culturing?
- 3) Describe the transmission ways, prevention.
- 4) Describe the treatment and required drugs for it. Explain the selection process of antibiotics for bacterial infection treatment.

### **Part 2**

#### **Types of questions:**

- 1) Characterize main pathogenic types of *Bacillus spp.*, their properties and differentiate the pathogenic factors in the development of pathological conditions.
- 2) Explain the rules of working with this microorganism? Culturing of pathogens, conditions.

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

### **List of drugs to repeat before final examination**

- |                               |                    |
|-------------------------------|--------------------|
| 1. Epinephrine                | Nifedipine         |
| 2. Norepinephrine             | Isoniazid          |
| 3. Enalapril                  | Streptomycin       |
| 4. Acetylsalicylic acid       | Metronidazole      |
| 5. Metoprolol                 | Ciprofloxacin      |
| 6. Losartan                   | 28. Azithromycin   |
| 7. Hydrochlorothiazide        | 29. Gliclazide     |
| 8. Insulin Actrapid           | 30. Fluconazole    |
| 9. Insulin NPH                | 31. Acyclovir      |
| 10. Fluticasone               | 32. Amphotericin B |
| 11. Salbutamol (Albuterol)    | 33. Rifampicin     |
| 12. Amoxicillin/clavulanate   | 34. Furazolidone   |
| 13. Cortisol (hydrocortisone) | 35. Co-trimoxazole |
| 14. Spironolactone            | 36. Pioglitazone   |
| 15. Metformin                 | 37. Morphine       |
| 16. Neostigmine               | 38. Diclofenac     |
| 17. Dexamethasone             | 39. Rivaroxaban    |
| 18. Heparin                   | 40. Nitroglycerin  |
| 19. Warfarin                  | 41. Doxycycline    |
| 20. Furosemide                | 42. Vancomycin     |
| 21. Ceftriaxone               | 43 Interferon      |
| 22. Prazosine                 |                    |

**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 3 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

<b>Evaluation</b>	<b>Criteria</b>	<b>Scale, points</b>
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 applicable), most key aspects identified and interpreted; 6. Correct use of professional terminology	50 - 69
Unsatisfactory (FX)	1. Most key aspects missed; 2. Lack of focus on question - no relevance and notable redundancy; 3. Some theoretical issues presented in some way; 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
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### 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” <i>(not taken into account when calculating GPA)</i>

### 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

3. Jawetz, Melnick & Adelberg's Medical microbiology. Geo F. Brooks, Karen C. Carroll, Janet S. Butel, Stephen A. Morse, Timothy A. Mietzner. 26<sup>th</sup> edition, 2013

**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. Computer program "Diamorph" - "Medical Microbiology" - atlas-guide to the bacteriology of mycology, protozoology and virology edited by Acad. Prof. Vorobyova A.A. <https://www.msmanuals.com/professional/clinical-pharmacology>