

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

**METHODICAL INSTRUCTIONS FOR PRACTICAL CLASSES**

MZiB2216 "Mechanisms of Defense and Disease (medical genetics, medical microbiology, general pharmacology)"  
**(10 credits)**  
**Spring semester, 2022-2023 academic year**

**Medical Genetics**

**Practical class 1-2**

**Topic:** Introduction to Medical Genetics. Chromosomal disorders.

**Content:** Classification of Hereditary diseases. Chromosomal mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of chromosomal disorders. Epidemiology of chromosomal disorders. Down syndrome as an example of autosomal chromosomal diseases (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology

**Maximum points – 3.6**

**Learning outcomes:**

1. draw mind map of hereditary diseases and explain principles of their classification;
2. identify clinical features of Down syndrome and explain its clinical variability and summarize clinical manifestation of autosomal chromosomal disorders;
3. explain genetic mutations as cause of Down syndrome and summarize their role in clinical variability of autosomal chromosomal disorders;
4. compare different diagnostic strategies for diagnosing Down syndrome and summarize principles of diagnosis of chromosomal disorders and discuss related ethical and legal issues;
5. compare and contrast different management strategies for Down syndrome and summarize principles of management of chromosomal disorders (prevention, treatment) and discuss related ethical and legal issues;
6. summarize epidemiologic data of chromosomal disorders on the example of Down syndrome;
7. calculate the risk of Down syndrome and summarize risk assessment strategy for chromosomal disorders;
8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with chromosomal disorders on the example of Down syndrome;
9. discuss impact of diagnosis of a genetic condition for the individual and the family;
10. demonstrate respect of patient's religious, cultural, social and ethical beliefs and understanding of how that might affect the decisions the patients make;
11. work with genetic databases (OMIM & etc);
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;

**Practical class 3**

**Topic:** Sex Chromosome disorders. Summary of chromosomal diseases.

**Content:** Classification of Hereditary diseases. Chromosomal mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of chromosomal disorders. Epidemiology of chromosomal disorders. Turner syndrome as an example of gonosomal chromosomal diseases (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology.

**Maximum points – 1.8**

**Learning outcomes:**

1. identify clinical features of Turner syndrome and explain its clinical variability and summarize clinical manifestation of gonosomal chromosomal disorders;
2. explain genetic mechanism of Turner syndrome and summarize their role in clinical variability of gonosomal chromosomal disorders;
3. compare and contrast different diagnostic strategies of Turner syndrome and summarize principles of diagnosis of gonosomal chromosomal disorders and discuss related ethical and legal issues;
4. compare and contrast different management strategies of Turner syndrome and summarize principles of management of gonosomal chromosomal disorders (prevention, treatment) and discuss related ethical and legal issues
5. summarize epidemiologic data of gonosomal chromosomal disorders on the example of Turner syndrome;
6. calculate the risk of Turner syndrome and summarize risk assessment strategy for gonosomal chromosomal disorders;
7. discuss a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with gonosomal chromosomal disorders on the example of Turner syndrome;
8. discuss impact of diagnosis of a genetic condition for the individual and the family;
9. demonstrate respect patient's religious, cultural, social and ethical beliefs and understanding of how that might affect the decisions the patients make
10. work with genetic databases (OMIM & etc).
11. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
12. complete mind map of hereditary diseases and discuss principles of their classification;
13. summarize genetic and medical aspects of chromosomal disorders: phenotypic manifestation, causes, mechanisms, epidemiology, principles and methods of prevention, diagnosis and management;

**Practical class 4**

**Topic:** Mendelian classic disorders: autosomal inheritance.

**Content:** Classifications, pathogenetic, pathogenetic mechanisms, epidemiology and management. Classification of classic Mendelian disorders. Gene mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of classic Mendelian disorders. Epidemiology of single gene disorders, prognosis. Cystic fibrosis as an example of an autosomal recessive disease (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology

**Maximum points – 1.8****Learning outcomes:**

1. draw mind map of single gene diseases (mendelain disorders) and explain principles of their classification;
2. identify clinical features of Cystic fibrosis and explain its clinical variability and summarize clinical manifestation of autosomal (dominant and recessive) Mendelian disorders;
3. explain mechanism of autosomal recessive mutations for Cystic fibrosis and summarize their role in clinical variability of it and of autosomal (dominant and recessive) monogenic disorders;
4. compare and contrast different diagnostic strategies of Cystic fibrosis and summarize principles of diagnosis of autosomal (dominant and recessive) monogenic disorders and discuss related ethical and legal issues;
5. compare and contrast different management strategies of Cystic fibrosis and summarize principles of management of autosomal (dominant and recessive) monogenic disorders (prevention, treatment) and discuss related ethical and legal issues;
6. summarize epidemiologic features of autosomal (dominant and recessive) monogenic disorders on the example of Cystic fibrosis;

7. calculate the risk of Cystic fibrosis and summarize risk assessment strategy for autosomal (dominant and recessive) monogenic disorders;
8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with autosomal (dominant and recessive) monogenic disorders on the example of Cystic fibrosis;
9. discuss impact of diagnosis of a genetic condition for the individual and the family;
10. demonstrate respect of patient's religious, cultural, social and ethical beliefs and understanding how that might affect the decisions the patients make;
11. work with genetic databases (OMIM & etc);
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
13. demonstrate understanding of genetic and medical aspects of chromosomal disorders on colloquium;

### **Practical class 5**

**Topic:** Mendelian classic disorders: autosomal inheritance.

**Content:** Classifications, pathogenetic mechanisms, epidemiology and management. Classification of classic Mendelian disorders. Gene mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of classic Mendelian disorders. Epidemiology of single gene disorders, prognosis. Cystic fibrosis as an example of an autosomal recessive disease (case study- continuation): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology

#### **Maximum points – 1.8**

#### **Learning outcomes:**

1. draw mind map of single gene diseases (Mendelian disorders) and explain principles of their classification;
2. identify clinical features of Cystic fibrosis and explain its clinical variability and summarize clinical manifestation of autosomal (dominant and recessive) Mendelian disorders;
3. explain mechanism of autosomal recessive mutations for Cystic fibrosis and summarize their role in clinical variability of it and of autosomal (dominant and recessive) monogenic disorders;
4. compare and contrast different diagnostic strategies of Cystic fibrosis and summarize principles of diagnosis of autosomal (dominant and recessive) monogenic disorders and discuss related ethical and legal issues;
5. compare and contrast different management strategies of Cystic fibrosis and summarize principles of management of autosomal (dominant and recessive) monogenic disorders (prevention, treatment) and discuss related ethical and legal issues;
6. summarize epidemiologic features of autosomal (dominant and recessive) monogenic disorders on the example of Cystic fibrosis;
7. calculate the risk of Cystic fibrosis and summarize risk assessment strategy for autosomal (dominant and recessive) monogenic disorders;
8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with autosomal (dominant and recessive) monogenic disorders on the example of Cystic fibrosis;
9. discuss impact of diagnosis of a genetic condition for the individual and the family;
10. demonstrate respect of patient's religious, cultural, social and ethical beliefs and understanding how that might affect the decisions the patients make;
11. work with genetic databases (OMIM & etc);
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;

### **Practical class 6**

**Topic:** Mendelian classic disorders: sex-linked inheritance.

**Content:** Classifications, pathogenetic mechanisms, epidemiology and management. Classification of Mendelian classic disorders. Gene mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of Mendelian classic disorders. Epidemiology of single gene disorders, prognosis. Hemophilia as an example of X-linked diseases (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology.

**Maximum points – 1.8**

**Learning outcomes:**

- 1. complete mind map of single gene diseases (Mendelian disorders) and explain principles of their classification;*
- 2. identify clinical features of Hemophilia and explain its clinical variability and summarize clinical manifestation of sex-linked Mendelian disorders;*
- 3. explain mechanism of genetic mutations in case of Hemophilia and summarize their role in clinical variability of sex-linked Mendelian disorders;*
- 4. compare and contrast different diagnostic strategies of Hemophilia and summarize principles of diagnosis of sex-linked Mendelian disorder and discuss related ethical and legal issues;*
- 5. compare and contrast different management strategies of Hemophilia and summarize principles of management of sex-linked Mendelian disorder (prevention, treatment) and discuss related ethical and legal issues;*
- 6. summarize epidemiologic data of sex-linked Mendelian disorders on the example of Hemophilia*
- 7. calculate the risk of Hemophilia and summarize risk assessment strategy for sex-linked Mendelian disorders;*
- 8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with sex-linked Mendelian disorders on the example of Hemophilia;*
- 9. discuss impact of diagnosis of a genetic condition on the individual and the family;*
- 10. demonstrate respect of patient's religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make*
- 11. work with genetic databases (OMIM & etc).*
- 12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;*

### **Practical class 7**

Colloquium “Introduction to Medical Genetics. Chromosomal disorders. Mendelian classic disorders”

**Topic:** Non-mendelian genetic disorders.

**Content:** Causes, classifications, pathogenetic mechanisms, epidemiology, diagnosis and management. Mitochondrial diseases. Genomic imprinting. Epigenetic of depression. Trinucleotide Repeat disorders. Huntington's Disease as an example of non-mendelian genetic disorders (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology. Summary of Monogenic diseases

**Maximum points – 16**

**Learning outcomes:**

- 1. draw mind map of non-mendelian genetic disorders and explain principles of their classification*
- 2. identify clinical features of Huntington's Disease and explain its clinical variability and summarize clinical manifestation of this disorders;*
- 3. explain mechanism of genetic mutations Huntington's Disease and summarize their role in clinical variability of non-mendelian genetic disorders;*
- 4. compare and contrast different diagnostic strategies of Huntington's Disease and summarize principles of diagnosis of non-mendelian genetic disorders and discuss related ethical and legal issues;*
- 5. compare and contrast different management strategies of Huntington's Disease and summarize principles of management of non-mendelian genetic disorders (prevention, treatment) and discuss related ethical and legal issues;*

6. summarize epidemiologic data of non-mendelian genetic disorders on the example of Huntington's Disease;
7. calculate the risk of Huntington's Disease and summarize risk assessment strategy for non-mendelian genetic disorders;
8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with non-mendelian genetic disorders on the example of Huntington's Disease;
9. discuss impact of diagnosis of a genetic condition on the individual and the family;
10. demonstrate respect of patient's religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make
11. work with genetic databases (OMIM & etc).
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
13. summarize genetic and medical aspects of monogenic disorders: phenotypic manifestation, causes, mechanisms, epidemiology, principles and methods of prevention, diagnosis and management.

### **Practical class 8**

**Topic:** Non-mendelian genetic disorders.

**Content:** Causes, classifications, pathogenetic mechanisms, epidemiology, diagnosis and management. Mitochondrial diseases. Genomic imprinting. Epigenetic of depression. Trinucleotide Repeat disorders. Huntington's Disease as an example of non-mendelian genetic disorders (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology. Summary of Monogenic diseases

**Maximum points – 1.8**

**Learning outcomes:**

1. draw mind map of non-mendelian genetic disorders and explain principles of their classification
2. identify clinical features of Huntington's Disease and explain its clinical variability and summarize clinical manifestation of this disorders;
3. explain mechanism of genetic mutations Huntington's Disease and summarize their role in clinical variability of non-mendelian genetic disorders;
4. compare and contrast different diagnostic strategies of Huntington's Disease and summarize principles of diagnosis of non-mendelian genetic disorders and discuss related ethical and legal issues;
5. compare and contrast different management strategies of Huntington's Disease and summarize principles of management of non-mendelian genetic disorders (prevention, treatment) and discuss related ethical and legal issues;
6. summarize epidemiologic data of non-mendelian genetic disorders on the example of Huntington's Disease;
7. calculate the risk of Huntington's Disease and summarize risk assessment strategy for non-mendelian genetic disorders;
8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with non-mendelian genetic disorders on the example of Huntington's Disease;
9. discuss impact of diagnosis of a genetic condition on the individual and the family;
10. demonstrate respect of patient's religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make
11. work with genetic databases (OMIM & etc).
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
13. summarize genetic and medical aspects of monogenic disorders: phenotypic manifestation, causes, mechanisms, epidemiology, principles and methods of prevention, diagnosis and management.

### **Practical class 9**

**Topic:** Fundamentals of the Population Genetics.

**Content:** Demographic characteristics, types of populations, marital structure of populations, genetic characteristics of the population. Hardy-Weinberg's law. Genetic burden of populations: concept and medical significance.

**Maximum points – 1.8**

**Learning outcomes:**

- 1. characterize population and genetic processes: mutations, selection, migration, and gene drift.*
- 2. explain patterns of the distribution of genes that make up the gene pool, including genes that determine hereditary human diseases and make links with disease cases discussed earlier;*
- 3. calculate distribution of genes and genotypes of disease in given genetic cases;*
- 4. justify the importance of studying hereditary diseases in human populations, its genetic diversity, identifying the frequencies of individual diseases and assessing the total load of hereditary human diseases.*
- 5. explain the phenomenon of person's genetic burden and discuss hereditary diseases as part of a genetic burden.*
- 6. discuss the importance of determination of the burden of hereditary diseases in human populations, the study of the magnitude and structure of the burden of hereditary diseases to determine the amount of medical, social and rehabilitation assistance to the population.*
- 7. demonstrate understanding of genetic and medical aspects of mendelian and non-mendelian genetic disorders on colloquium.*

### **Practical class 10**

**Topic:** Fundamentals of the Population Genetics.

**Content:** Demographic characteristics, types of populations, marital structure of populations, genetic characteristics of the population. Hardy-Weinberg's law. Genetic burden of populations: concept and medical significance.

**Maximum points – 1.8**

**Learning outcomes:**

- 1. Characterize population and genetic processes: mutations, selection, migration, and gene drift.*
- 2. Explain patterns of the distribution of genes that make up the gene pool, including genes that determine hereditary human diseases and make links with disease cases discussed earlier;*
- 3. Know how to apply the Hardy-Weinberg equilibrium and solve problems concerning genotype and allele frequencies*
- 4. Interpret scenarios about factors responsible for genetic variation in/ among populations*
- 5. Justify the importance of studying hereditary diseases in human populations, its genetic diversity, identifying the frequencies of individual diseases and assessing the total load of hereditary human diseases.*
- 6. Explain the phenomenon of person's genetic burden and discuss hereditary diseases as part of a genetic burden.*
- 7. Discuss the importance of determination of the burden of hereditary diseases in human populations, the study of the magnitude and structure of the burden of hereditary diseases to determine the amount of medical, social and rehabilitation assistance to the population.*

### **Practical class 11-12**

**Topic:** Polygenic multifactorial disorders.

**Content:** characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of polygenic diseases. Epidemiology of polygenic diseases. Diabetes mellitus as an example of polygenic multifactorial disorders (case study): signs and symptoms, classification, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology. How Insulin work. The role of Glucose. Prediabetes: causes and risk groups. Differences Diabetes 1 type and Diabetes 2 type. Complications.

**Maximum points – 3.6**

1. *continue work on mind map of hereditary diseases and explain principles of classification of polygenic disorders;*
2. *identify clinical features of Diabetes mellitus and explain its clinical variability and summarize clinical manifestation of polygenic disorders;*
3. *explain mechanism of polygenic inheritance and summarize their role in clinical variability of polygenic disorders;*
4. *compare and contrast different strategies in genetic profiling of Diabetes mellitus and summarize principles of diagnosis and genetic screening of polygenic disorders and discuss related ethical and legal issues;*
5. *compare and contrast different management strategies of Diabetes mellitus and summarize principles of management of polygenic disorders (prevention, treatment) and discuss related ethical and legal issues;*
6. *summarize epidemiologic data of polygenic disorders on the example of Diabetes mellitus;*
7. *calculate the risk of Diabetes mellitus and summarize risk assessment strategies for polygenic disorders;*
8. *identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with polygenic disorders on the example of Diabetes mellitus;*
9. *discuss impact of diagnosis of a genetic condition on the individual and the family;*
10. *demonstrate respect of patient's religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make*
11. *work with genetic databases (OMIM & etc).*
12. *demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;*
13. *summarize genetic and medical aspects of monogenic disorders: phenotypic manifestation, causes, mechanisms, epidemiology, principles and methods of prevention, diagnosis and management;*

### **Practical class 13**

**Topic:** Cancer Genetics and Genomics.

**Content:** Cancer genes. Hereditary cancer syndromes. Familial occurrence of cancer. Sporadic cancer and genetic bases. Genetic technology in cancer prevention, diagnosis and therapy. Breast cancer (case study): causes and genetic mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology.

**Maximum points – 1.8**

**Learning outcomes:**

1. *continue work on mind map of hereditary diseases and explain principles of their classification*
2. *identify clinical features of breast cancer and explain its lifetime prevalence, genetic variability and summarize data on hereditary cancer syndromes and syndromes with familial cancer;*
3. *explain genetic mechanisms of oncogenesis on breast cancer example and summarize their role in clinical variability of hereditary cancer syndromes and syndromes with familial cancer;*
4. *compare and contrast different diagnostic strategies of breast cancer and summarize principles of diagnosis of hereditary cancer syndromes and syndromes with familial cancer and discuss related ethical and legal issues;*
5. *compare and contrast different management strategies of breast cancer and summarize principles of management of hereditary cancer syndromes and syndromes with familial cancer (prevention, treatment) and discuss related ethical and legal issues;*
6. *summarize epidemiologic data of hereditary cancer syndromes and syndromes with familial cancer on the example of breast cancer;*
7. *calculate the risk of breast cancer and summarize risk assessment strategies for hereditary cancer syndromes and syndromes with familial cancer;*

8. *identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with hereditary cancer syndromes and syndromes with familial cancer on the example of breast cancer*
9. *discuss impact of diagnosis of hereditary cancer syndromes and syndromes with familial cancer on the individual and the family;*
10. *demonstrate respect of patient's religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make*
11. *work with genetic databases (OMIM & etc).*
12. *demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;*
13. *summarize genetic and medical aspects of monogenic disorders: phenotypic manifestation, causes, mechanisms, epidemiology, principles and methods of prevention, diagnosis and management;*

#### **Practical class 14**

**Topic:** Polygenic disorders: developmental malformation.

**Content:** Classification, cause, mechanisms, prevalence rates, phenotypic manifestation, clinical significance. Neural tube defects (NTDs) as example (case study): definition, types, causes, mechanisms, diagnosis, prevention, epidemiology and management.

**Maximum points – 1.8**

**Learning outcomes:**

1. *continue work on mind map of hereditary diseases and explain principles of their classification*
2. *identify clinical features of NTDs and explain its clinical variability and summarize clinical manifestation of developmental malformation in general;*
3. *explain mechanism of genetic causes of NTDs and summarize their role in clinical variability of developmental malformation in general;*
4. *compare and contrast different diagnostic strategies of NTDs and summarize principles of diagnosis of developmental malformation in general and discuss related ethical and legal issues;*
5. *compare and contrast different management strategies of NTDs and summarize principles of management of developmental malformation in general (prevention, treatment) and discuss related ethical and legal issues;*
6. *summarize epidemiologic data of developmental malformation in general on the example of NTDs;*
7. *calculate the risk of NTDs and summarize risk assessment strategy for developmental malformation;*
8. *identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with developmental malformation on the example of NTDs;*
9. *discuss impact of diagnosis of developmental malformation on the individual and the family;*
10. *demonstrate respect of patient's religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make;*
11. *work with genetic databases (OMIM & etc);*
12. *demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;*

#### **Practical class 15**

Colloquium “Non-mendelian classic disorders. Population genetics. Polygenic multifactorial disorders”

**Topic:** Polygenic disorders: developmental malformation.

**Content:** Classification, cause, mechanisms, prevalence rates, phenotypic manifestation, clinical significance. Neural tube defects (NTDs) as example (case study-continuation): definition, types, causes, mechanisms, diagnosis, prevention, epidemiology and management.

**Maximum points – 16**

**Learning outcomes:**

1. *continue work on mind map of hereditary diseases and explain principles of their classification*



2. *identify clinical features of NTDs and explain its clinical variability and summarize clinical manifestation of developmental malformation in general;*
3. *explain mechanism of genetic causes of NTDs and summarize their role in clinical variability of developmental malformation in general;*
4. *compare and contrast different diagnostic strategies of NTDs and summarize principles of diagnosis of developmental malformation in general and discuss related ethical and legal issues;*
5. *compare and contrast different management strategies of NTDs and summarize principles of management of developmental malformation in general (prevention, treatment) and discuss related ethical and legal issues;*
6. *summarize epidemiologic data of developmental malformation in general on the example of NTDs;*
7. *calculate the risk of NTDs and summarize risk assessment strategy for developmental malformation;*
8. *identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with developmental malformation on the example of NTDs;*
9. *discuss impact of diagnosis of developmental malformation on the individual and the family;*
10. *demonstrate respect of patient's religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make*
11. *work with genetic databases (OMIM & etc).*
12. *demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;*
13. *summarize genetic and medical aspects of developmental genetics: phenotypic manifestation of developmental malformation, causes, mechanisms, epidemiology, principles and methods of prevention, diagnosis and management*
14. *demonstrate understanding of genetic and medical aspects of population genetics and polygenic multifactorial disorders on colloquium.*

## **METHODICAL INSTRUCTIONS FOR PRACTICAL CLASSES**

**Aim:** to enforce understanding of pathogenesis, methods of diagnosis and management of genetically determined and hereditary diseases, develop problem solving, team-working and self-learning skills.

### **Learning outcomes:**

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. *understand the biochemical processes in the main pathological conditions and genetically determined diseases.*
3. *interpret the results of specific molecular genetic diagnostic methods*
4. *understand the role of relevant risk factors of diseases for decision-making with a view to their prevention.*
5. *integrate knowledge on human genetics for the purposes of diagnosis and personalized treatment of human pathology*
6. *demonstrate the ability to identify learning gaps and create strategies to enhance one's own knowledge and skills.*
7. *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.*

### **Work schedule**

1. Familiarize yourself with the basic and additional literature, use textbooks, the syllabus and present directions, Internet resources to prepare for seminars.
2. Be prepared for class and participate actively on case-discussion and problem-solving group activities.
3. Use the examples (in this number cases and your own experience studied before) for illustration of theoretic material.
4. Use different tools for studying, discussion and visualization of thoughts - drawing, mind maps, 3d-modelling.
5. Use the group work with cases for the development of teamwork skills, communication, problem solving and self-studying.

## **Medical Microbiology**

### **Practical class 1**

**Topic:** 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.

**Maximum points – 1.8**

#### **Learning Outcomes:**

1. *characterize main types of gram-positive and gram-negative cocci, their properties,*
2. *explain their role and pathogenesis of the development of pathological conditions,*
3. *justify the principles of laboratory diagnosis and prevention and treatment of the diseases caused by them*
4. *model isolation of a pure microbe culture and interpret the result*

### **Practical class 2**

**Topic:** 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.

**Maximum points – 1.8**

#### **Learning Outcomes:**

1. *characterize microorganisms of the intestinal group of bacteria*
2. *differentiate the properties of Escherichia and Shigella and explain their role in the development of pathological conditions, pathogenesis, caused diseases,*
3. *justify features of microbiological diagnosis in connection with the pathogenesis of diseases,*
4. *justify principles of prevention and treatment*
5. *model isolation of a pure microbe culture and interpret the result*

### **Practical class 3**

**Topic:** 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.

**Maximum points – 1.8**

#### **Learning Outcomes:**

1. *describe properties of Salmonella, and explain their role in the development of pathological conditions, pathogenesis, caused diseases,*
2. *justify features of microbiological diagnosis in connection with the pathogenesis of diseases,*
3. *justify principles of prevention and treatment*
4. *argue the role of campylo- and helicobacter in the development of pathological conditions*

#### **Practical class 4**

**Topic:** 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.

**Maximum points – 1.8**

##### **Learning Outcomes:**

- 1. differentiate causative agents of zoonotic infections, their properties, explain pathogenesis of the development of diseases,*
- 2. justify features of microbiological diagnosis in connection with the pathogenesis of diseases,*
- 3. justify principles of prevention and treatment*
- 4. explain the concept of quarantine infections and the rules of the anti-epidemic regime in the occurrence and development of anthrax and plague*
- 5. model serological diagnosis of anthrax and brucellosis with interpretation of the results*

#### **Practical class 5**

**Topic:** 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.

**Maximum points – 1.8**

##### **Learning Outcomes:**

- 1. differentiate causative agents of diphtheria and pertussis, their properties, explain pathogenesis of the development of diseases,*
- 2. justify features of microbiological diagnosis in connection with the pathogenesis of diseases,*
- 3. justify principles of prevention and treatment*
- 4. explain a concept of toxinemic infections*

#### **Practical class 6**

**Topic:** 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.

**Maximum points – 1.8**

##### **Learning Outcomes:**

- 1. differentiate causative agent of tuberculosis and leprosy, its properties, explain pathogenesis of the development of the disease,*
- 2. justify features of microbiological diagnosis in connection with the pathogenesis of the diseases,*
- 3. justify principles of prevention and treatment*
- 4. explain vaccination rules for the prevention of tuberculosis*
- 5. discuss general principles of DOTS treatment of tuberculosis*

#### **Practical class 7**

**Topic:** 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.

**Maximum points – 1.8**

##### **Learning Outcomes:**

- 1. differentiate causative agent of sexually transmitted diseases, its properties, explain pathogenesis of the development of the disease,*
- 2. justify features of microbiological diagnosis in connection with the pathogenesis of the disease,*

### *3. justify principles of prevention and treatment*

#### **Practical class 8**

**Topic:** The causative agents of anaerobic infections. Algorithm for laboratory diagnosis of anaerobic infections. Features of microbiological diagnosis in connection 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.

**Maximum points – 1.8**

**Learning Outcomes:**

- 1. differentiate causative agent of anaerobic infections, its properties, explain pathogenesis of the development of the disease,*
- 2. justify features of microbiological diagnosis in connection with the pathogenesis of the disease,*
- 3. justify principles of prevention and treatment*

#### **Practical class 9**

**Topic:** Adenoviruses. Poxviruses. Rhabdoviruses. Role in human pathology. The principles of treatment. Prevention.

**Maximum points – 1.8**

**Learning Outcomes:**

- 1. differentiate causative agents of respiratory infections, their properties,*
- 2. explain their role in the development of pathological conditions,*
- 3. justify principles of laboratory diagnosis and prevention and treatment of diseases caused by them*
- 4. interpret the results of laboratory diagnosis.*

#### **Practical lesson 10**

**Topic:** Orthomyxoviruses (influenza virus). Paramyxoviruses (viruses of parainfluenza, mumps, measles, respiratory syncytial virus). Statement of RGA, RTGA, RTGA in paired sera. Interpretation of the results.

**Maximum points – 1.8**

**Learning Outcomes:**

- 5. differentiate causative agents of respiratory infections, their properties,*
- 6. explain their role in the development of pathological conditions,*
- 7. justify principles of laboratory diagnosis and prevention and treatment of diseases caused by them*
- 8. interpret the results of laboratory diagnosis.*

#### **Practical class 11**

**Topic:** 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.

**Maximum points – 1.8**

**Learning Outcomes:**

- 1. differentiate causative agents of poliomyelitis, rubella, their properties,*
- 2. explain their role in the development of pathological conditions,*
- 3. justify principles of laboratory diagnosis and prevention and treatment of the diseases caused by them*
- 4. interpret color test results*

#### **Practical class 12**

**Topic:** 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

**Maximum points – 1.8**

**Learning Outcomes:**

1. *differentiate causative agent of HIV infection, its properties,*
2. *explain their role in the development of AIDS,*
3. *justify principles of laboratory diagnosis and prevention and treatment*
4. *describe ELISA test and interpretation of results*

### **Practical class 13**

**Topic:** Hepatitis A, B, C. viruses. Treatment principles, prevention. Herpes viruses (alpha beta, gamma herpes viruses). Principles of treatment, prevention.

**Maximum points – 1.8**

**Learning Outcomes:**

1. *differentiate causative agent of hepatitis and herpetic infection, its properties,*
2. *explain their role in the development of AIDS,*
3. *justify principles of laboratory diagnosis and prevention and treatment*

### **Practical class 14**

**Topic:** Fungal infections or mycoses. Candidiasis, Cryptococcosis, Aspergillosis, Blastomycosis. Laboratory diagnostic, treatment principles, prevention.

**Maximum points – 1.8**

**Learning Outcomes:**

1. *differentiate causative agent of mycoses, its properties,*
2. *explain their role in the development of pathological conditions,*
3. *justify principles of laboratory diagnosis and prevention and treatment*

### **Practical class 15**

**Topic:** Nosocomial diseases. Classification, risks, prevention, clinical cases

**Maximum points – 1.8**

**Learning Outcomes:**

1. *explain the role of popular nosocomial infections in the development of pathological conditions,*
2. *justify features of microbiological diagnosis in connection with the pathogenesis of the disease,*
3. *justify principles of prevention and treatment*

## **METHODICAL INSTRUCTIONS FOR PRACTICAL CLASSES**

**Aim:** to enforce understanding of the role of microorganisms in human infectious pathology, the use of microbiological methods in the diagnosis of diseases, develop problem solving, team-working and self-learning skills.

**Learning outcomes:**

1. *apply knowledge of the infectious process and its features in various types of human pathogens, apply knowledge of immunodiagnosics of infectious diseases, demonstrate an understanding of the principles of infection control and biosafety*
2. *understand the role of relevant risk factors of diseases for decision-making with a view to their prevention.*
3. *integrate knowledge on the interaction of micro and macro-organism for the purposes of diagnosis and personalized treatment of human pathology*
4. *demonstrate the ability to identify learning gaps and create strategies to enhance one's own knowledge and skills.*
5. *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*

## **Work schedule**

1. Familiarize yourself with the basic and additional literature, use textbooks, the syllabus and present directions, Internet resources to prepare for seminars.
2. Be prepared for class and participate actively on case-discussion and problem-solving group activities.
3. Use the examples (in this number cases and your own experience studied before) for illustration of theoretic material.
4. Use different tools for studying, discussion and visualization of thoughts - drawing, mind maps, 3d-modelling.
5. Use the group work with cases for the development of teamwork skills, communication, problem solving and self-studying.

## **General Pharmacology**

### **Practical class 1**

**Topic:** Introduction to Pharmacology. The value of the subject. Dosage Forms. INN, trade names. Medicinal dosage forms. Drug prescription.

**Maximum points – 2.4**

#### **Learning outcomes:**

1. *Explain the purpose of the science of pharmacology and its basic terms.*
2. *List the basic dosage forms.*
3. *Explain the principles of naming drugs (Chemical names, international nonproprietary name, trade names, original and generic)*
4. *Write a prescription for the drug.*

### **Practical class 2**

**Topic:** Pharmacokinetics. Principles of interaction of human bodies with the drugs. Absorption, distribution, biotransformation and excretion of chemicals. Effects of impaired organ functions on pharmacokinetics.

**Maximum points – 2.4**

#### **Learning outcomes:**

1. *Explain terms: “pharmacokinetics, absorption, distribution, biotransformation, excretion”*
2. *Recognize the routes of drugs inside human bodies*
3. *Apply this knowledge when describing a drug.*

### **Practical class 3**

**Topic:** Pharmacodynamics. Principles of interaction of drugs with human bodies. Different mechanisms of action – agonism and antagonism to different types and subtypes of receptors, inhibition of enzymes, blocking or opening of channels.

**Maximum points – 2.4**

#### **Learning Outcomes:**

1. *Explain terms: “pharmacodynamics, receptor, channel, enzyme, agonist, antagonist, partial agonist, inhibitor, channel blocker, channel transporter”*
2. *Understand the mechanism of drug action on chemical and anatomical basis.*
3. *Apply this knowledge when describing a drug*

### **Practical class 4**

**Topic:** PNS. Cholinergic drugs. Acetylcholine, its function in healthy human body. M and N cholinergic receptors, different subtypes. Cholinomimetics. Cholinesterase inhibitors.

**Maximum points – 2.4**

#### **Learning Outcomes:**

1. *Explain the functions and location of  $M_1$ ,  $M_2$ ,  $M_3$ ,  $N_N$ ,  $N_M$  receptors throughout the human body.*
2. *Describe action of cholinesterase inhibitors.*
3. *Demonstrate, how selectivity to different types of receptors linked to drug action.*

4. Characterize (indications, contraindications, side effects) of this drugs: *Pilocarpine, Physostigmine, Galantamine, Neostigmine, Nicotine, Cytisine.*

#### **Practical class 5**

**Topic:** PNS. Cholinergic drugs. Cholinoblockers. Cholinesterase reactivators.

**Maximum points – 2.4**

**Learning Outcomes:**

1. Explain the main effects of cholinoblockers.
2. Describe action of cholinoblockers and cholinesterase reactivators.
3. Demonstrate, how selectivity to different types of receptors linked to drug action.
4. Characterize (indications, contraindications, side effects) of this drugs: *Pipekuronium, Succinylcholine, Atropine, Solifenacin. Hyoscine, Piridoxim*

#### **Practical class 6**

**Topic:** PNS. Adrenergic drugs. Noradrenaline and adrenaline (Norepinephrine and epinephrine), their functions in healthy human body. Alfa and beta adrenoreceptors, different subtypes. Adrenomimetics.

**Maximum points – 2.4**

**Learning Outcomes:**

1. Describe the functions and location of  $\alpha_1$ ,  $\alpha_2$ ,  $\beta_1$ ,  $\beta_2$ ,  $\beta_3$  receptor subtypes throughout the human body.
2. Describe action of adrenomimetics.
3. Demonstrate, how selectivity to different types of receptors linked to drug action.
4. Characterize (indications, contraindications, side effects) this drugs: *Adrenaline (epinephrine), Phenylephrine, Naphazoline, Ephedrine, Clonidine, Dobutamine, Salbutamol, Salmeterol, Isoprenaline (historical)*

#### **Practical class 7**

**Topic:** PNS. Adrenergic drugs. Adrenoblockers, their functions in healthy human body. Alfa and beta adrenoreceptors blockers, different subtypes. Sympatholytics.

**Maximum points – 2.4**

**Learning Outcomes:**

1. Describe action of adrenoblockers.
2. Demonstrate, how selectivity to different types of receptors linked to drug action.
3. Characterize (indications, contraindications, side effects) this drugs: *Phentolamine (historical), Yohimbine, Prazosin / Doxazosin, Propranolol, Metoprolol, Labetalol, Carvedilol, Atenolol.*

#### **Practical class 8**

**Topic:** Antianginal drugs. Antihypertensive drugs. Diuretics, Ca channel blockers, Nitrates, ACEI

**Maximum points – 2.4**

**Learning Outcomes:**

1. Explain mechanisms of cardiac ischemia.
2. Explain function of RAAS (renin-angiotensin-aldosterone system).
3. Explain mechanisms of regulation of water-salt balance.
4. Compare different antianginal drugs.
5. Characterize (indications, contraindications, side effects) these drugs: *Nitroglycerin, Isosorbite dinitrate, verapamil, alpha-blockers (repeat), Beta-blockers (repeat), captopril, enalapril, losartan, nifedipine, amlodipine, clonidine, moxonidine, furosemide, hydrochlorothiazide, indapamide, spironolactone.*

#### **Practical class 9**

**Topic:** Pharmacology of the hematopoietic system and hemostasis. Preparations for the treatment of anemia. Coagulation disorders. Drugs, enhancing drugs and reducing coagulation. Drugs, increasing and reducing platelet aggregation.

**Maximum points – 2.4**

**Learning Outcomes:**

1. Compare and define different causes of anemia
2. List indications for antiagregant (antiplatelet) ant anticoagulant therapy
3. Explain mechanism of development of atherosclerosis.
4. Characterize (indications, contraindications, side effects) these drugs: iron supplements, folic acid and B12 preparation, ASA, clopidogrel, dabigatran, warfarin, rivaroxaban, menadione, aminocaproic acid, heparins, alteplasa, urokinase

**Practical class 10**

**Topic:** Pharmacology of ES. Diabetes melitus.

**Maximum points – 2.4**

**Learning Outcomes:**

1. Compare mechanisms of development of type I and type II diabetes melitus.
2. Explain principal insulin replacement therapy, its principles.
3. Describe drugs, used in in treatment of type II diabetes: Insulin secretagogues. Insulin sensitizers. Agents acting on the absorption and excretion of glucose.
4. Tell the function of glucagon and amylin
5. Characterize (indications, contraindications, side effects) these drugs: insulins, metformin, glibenclamide, repaglinide, pioglitazone, canagliflozin, liraglutide, sitagliptin

**Practical class 11**

**Topic:** Anti-inflammatory drugs. Nonsteroidal anti-inflammatory drugs. Steroidal anti-inflammatory drugs.

**Maximum points – 2.4**

**Learning Outcomes:**

1. Compare the functions and location of COX-1 and COX-2 enzyme subtypes throughout the human body.
2. List hormones f adrenal cortex
3. Characterize (indications, contraindications, side effects) these drugs: aspirin, diclofenac, ibuprofen, celecoxib, meloxicam prednisolone, dexamethasone, fludrocortisone.

**Practical class 12**

**Topic:** Opioid system. Opioid agonists and antagonists. Drug addiction

**Maximum points – 2.4**

**Learning Outcomes:**

1. Explain functions of nociceptive and antinociceptive system.
2. Tell about opioid receptors, their agonists and antagonists.
3. Give definitions to “abuse”, “addiction”, “tolerance”, “dependence”, “withdrawal”
4. Compare physical and psychological dependence
5. Characterize (indications, contraindications, side effects) these drugs: morphine, fentanyl, tramadol, buprenorphine, naloxone.

**Practical class 13**

**Topic:** Antibiotics. Principles of antimicrobial therapy. Mechanisms of formation, prevention and overcoming of resistance. Beta-lactams, Macrolides, Tetracyclines, Peptide antibiotics. Aminoglycosides.

**Maximum points – 2.4**

**Learning Outcomes:**

1. Explain the difference between gram-positive and gram-negative bacteria
2. Define fungi, chlamydia, mycoplasma, viruses.
3. Explain mechanisms of development of resistance
4. List methods of overcoming resistance
5. Characterize (indications, contraindications, side effects) this drugs: Penicillin, amoxicillin, oxacillin, cefazolin, cefuroxime, ceftriaxone cefepime, Ceftaroline, imipenem, aztreonam, clindamycin,



*erythromycin, azithromycin, clarithromycin, Streptomycin, gentamicin, doxycycline, Tigecycline, chloramphenicol, vancomycin,*

#### **Practical class 14**

**Topic:** Antibiotics. Nitroimidazoles and nitrofurans. fluoroquinolones. Linezolid. Sulfonamides. Trimethoprim.TB.

**Maximum points – 2.4**

#### **Learning Outcomes:**

1. *Explain the difference between antibiotics and synthetic antituberculous drugs*
2. *Compare different sulfonamide drugs*
3. *Explain mechanisms of action of combined drugs (trimethoprim)*
4. *Characterize (indications, contraindications, side effects) these drugs: metronidazole, furazolidone, nitroksolin, ciprofloxacin, linezolid, Sulfametoksazol, trimethoprim, isoniazid, pyrazinamide, ethambutol, rifampicin, ethionamide, streptomycin, PASA*

#### **Practical class 15**

**Topic:** Antiviral drugs. Treatment of HIV infection. Antifungals

**Maximum points – 2.4**

#### **Learning Outcomes:**

1. *Explain mechanisms of action of antiviral drugs.*
2. *Compare the different drugs against HIV.*
3. *Define antifungal for dermatomycosis and systemic mycosis.*
4. *Characterize (indications, contraindications, side effects) these drugs: acyclovir; rimantadine, ribavirin, sofosbuvir, interferons, amphotericin B, ketoconazole, fluconazole, caspofungin*

### **METHODICAL INSTRUCTIONS FOR PRACTICAL CLASSES**

**Aim:** This course is an introduction to pharmacology based on evidence-based medicine and placebo-controlled clinical trials. The course gives students a basic understanding of modern pharmacology and gives a broad overview of the relationship between the basic concepts in general biology (including cell transport, biochemistry and metabolism) and the drugs that affect them. The principles and mechanisms of the action of drugs in a clinical context, as well as at the cellular level are considered, then this knowledge is integrated into a single system. The concepts of anatomy, molecular biology and physiology are illustrated by medical examples to engage students in analytical thinking and to stimulate independent as well as joint work on educational material.

#### **Learning outcomes:**

1. *Discuss the principles of modern pharmacology based on the current achievements of science;*
2. *write a prescription for a drug;*
3. *apply the principles of pharmacokinetics in the work (absorption, distribution, biotransformation and excretion of drugs;*
4. *apply the principles of pharmacodynamics (mechanisms of action of drugs at the molecular level);*
5. *list about the main groups of drugs, their mechanisms of action, indications and contraindications for their use.*
6. *describe of unwanted and adverse drug reactions,*
7. *use the principles of evidence-based pharmacology and evidence-based medicine, justify the use of a drug from the perspective of evidence-based medicine;*
8. *apply the basics of medical international terminology, from the field of pharmacology;*
9. *integrate knowledge of anatomy, physiology and biochemistry to explain the mechanisms of action of drugs;*
10. *independently find, analyze and summarize educational and scientific information in relation to situations related to the course content;*

### Work schedule

1. Familiarize yourself with the basic and additional literature, use textbooks, the syllabus and present directions, Internet resources to prepare for seminars.
2. Be prepared for class and participate actively on case-discussion and problem solving group activities.
3. Use the examples (in this number cases and your own experience studied before) for illustration of theoretic material.
4. Use different tools for studying, discussion and visualisation of thoughts - drawing, mind maps, 3d-modelling.
5. Use the group work with cases for the development of teamwork skills, communication, problem solving and self-studying.

### Scale of response quality

<b>Evaluation</b>	<b>Criteria</b>	<b>Scale, points</b>
Excellent	<ol style="list-style-type: none"><li>1. All key aspects included and presented logically;</li><li>2. High accuracy (relevance, without redundancy) and consistent focus on question;</li><li>3. Excellent integration of theoretical issues;</li><li>4. Provision of relevant examples;</li><li>5. In-depth analysis and theoretical justification of given problem (if applicable), all key aspects identified and interpreted;</li><li>6. Fluency in use of professional terminology</li></ol>	90 - 100
Good	<ol style="list-style-type: none"><li>1. All key aspects included and presented logically;</li><li>2. Consistent focus on question with satisfactory accuracy, and relevance, and/or some redundancy;</li><li>3. Satisfactory integration of theoretical issues;</li><li>4. Lack of examples;</li><li>5. Satisfactory analysis and theoretical justification of given problem (if applicable), most key aspects identified and interpreted;</li><li>6. Correct use of professional terminology</li></ol>	75 - 89
Satisfactory	<ol style="list-style-type: none"><li>1. Most key aspects included;</li><li>2. Satisfactory focus on question - some lapses of relevance and/or noticeably redundancy;</li><li>3. Theoretical issues presented without noticeably integration;</li><li>4. Provision of unsuccessful examples or no examples;</li><li>5. Some analysis and theoretical justification of given problem (if applicable), most key aspects identified and interpreted;</li><li>6. Correct use of professional terminology</li></ol>	50 - 70
Unsatisfactory (FX)	<ol style="list-style-type: none"><li>1. Most key aspects missed;</li><li>2. Lack of focus on question - no relevance and notable redundancy;</li><li>3. Some theoretical issues presented in someway;</li><li>4. No or irrelevant examples;</li><li>5. Some analysis and theoretical justification of a given problem (if applicable), most key aspects missed;</li><li>6. Lapses in use of professional terminology</li></ol>	25 - 49

Failed	<ol style="list-style-type: none"> <li>1. Most or all key aspects missed;</li> <li>2. No focus on question, irrelevant information;</li> <li>3. Theoretical issues missed or superficial;</li> <li>4. No or irrelevant examples;</li> <li>5. No analysis and no theoretical justification of a given problem (if applicable), most key aspects missed;</li> <li>6. Lapses in use of professional terminology</li> </ol>	0-24
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### **TEAMWORK GUIDELINES**

The medical profession involves working in multidisciplinary teams, so these skills are identified as key in the competence of the doctor and other health professionals in all countries.

Therefore, group work is included as an essential component in the practical exercises of our course. In addition, it aims to provide a safe environment in which you can try out new ideas and practices and acquire relevant group skills. These can be tasks for performance in pairs, triples or small groups of 4-6 people (work with cases, tasks of the ISW, etc.).

When you are working on a project or task in a team, you have the opportunity to use the various strengths of the group members to create a wider and better project or task than if you were working independently.

Group training means you need to share your knowledge and ideas with other students. There are two benefits to this: you need to think carefully about your own ideas in order to explain them to others, and you expand your own understanding, taking into account the knowledge and ideas of others.

#### **Interpersonal Communication and Discussion**

Take some time to chat and get to know each of your group mates. The better you know each other and the more convenient you communicate, the more effective you can work together.

Create a culture of mutual respect in your group. You probably had little choice or no choice at all when forming training groups and small teams in the classroom. Therefore, you will have to learn to overcome the differences between people. In addition, you will not have the opportunity to choose employees in the workplace, and at work, you will experience much greater pressure to be a productive member of the team.

For effective communication and discussion in a team: you should not be shy to express your opinion and it is important to feel that these opinions will be heard; it is necessary to feel that all members of the group make a feasible contribution to solving problems, observing agreed rules and plans, performing work efficiently and on time; it is important to know that everyone's feelings are taken into account by team members, but the goals and objectives of the group are not compromised, in favor of the whims or desires of individual members;

Try to express your opinion and listen to others. There is nothing wrong with disagreeing with your classmates, no matter how confident they are. When you disagree, be constructive and focus on the problem, not the person. Similarly, when someone disagrees with you, respect what he says and the risk that he takes upon himself to express his opinion. Try to find a way that everyone can agree with, and this is not necessarily the opinion of the loudest or smartest member of the team. Below we provide some examples of constructive and destructive group behavior<sup>1</sup>:

Constructive group behavior - a person who:

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<sup>1</sup> adapted from Brunt (1993):

<https://tle.wisc.edu/solutions/engagement/constructive-and-destructive-groupbehaviors>

*Unites* - interest in the views and opinions of others and willingness to adapt to interest

*Clarifies* - clearly defines the problems for the group by listening, summarizing, focusing the discussion

*Inspires* - encourages the group, stimulates participation and progress

*Harmonizes* - stimulates group unity and teamwork. For example, uses humor as a relaxation after difficult situations.

*Take the risk* - willingness to take risks at the expense of oneself for the success of the group or project

*Manages the process* - organizes a group on the issues of the process: for example, plan, schedule, timeline, topic, solution methods, and use of information

Destructive group behavior:

*Domination* - takes a lot of time expressing your opinion and views. Trying to take control by capturing energy, time, etc.

*Fussiness* - hastens the group to move quickly before the task is completed. Impatient in listening to other opinions and working together.

*Suspension* - removes itself from a discussion or decision. Opt out

*Ignoring* - does not respect or belittle the ideas and suggestions of the team or individuals. An extreme manifestation of ignoring is an insult in the form of ridicule.

*Distraction* - excessive talkativeness, tells stories and leads groups away from the goal

*Blocking* - prevents group progress by denying all ideas and suggestions. "It will not work because ..."

Effective group work does not arise by itself. A conscious and planned effort is needed, and since many people participate in it, one cannot rely on memory; need to make notes. The following steps will help you and your team work together effectively.

1. *Define clear objectives.* At each stage, you should try to coordinate the tasks. They include a timeline for the project, as well as more specific tasks (such as "agree on an approach to the task before Friday"). Each meeting or discussion should also begin with a specific goal (for example, make a list of tasks that need to be completed). Tasks should be broken down into smaller parts and planned. Sometimes one part cannot be started until the other part is finished, so you may need to draw a simple temporary map.

- discuss the resources that you have and those that you will need to find.
- formulate the desired result.
- consider how you know when you did it well enough?
- split tasks between the team and
- set deadlines for subtasks and time for future meetings.

2. *Set the basic rules.* Discussions can become erratic and can prevent more modest group members from participating if you do not have rules to stimulate discussion, resolve disagreements, and make decisions without repetition. Set the rules from the start and change them as needed. For example: an interesting rule that was developed by one group - anyone who missed a meeting would buy the rest of the group coffee in a coffee shop. No one ever missed a meeting after that.

3. *Communicate effectively.* Make sure you regularly communicate with group members. Try to be clear and positive in what you say without repeating.

4. *Find consensus.* People work together most effectively when they work towards a goal with which they have agreed. Make sure everyone has their own opinion, even if you need time to get more participants to say something. Make sure you listen to everyone's ideas and then try to come to an agreement that everyone shares and everyone has contributed.

5. *Define the roles.* Divide the work that needs to be done into separate tasks, for which you can use the strengths of individual team members. Define roles for both fulfilling your tasks and for meetings / discussions (for example, Arani is responsible for summarizing the discussions, Joseph is for everyone to express their opinions and make decisions, etc.).

Examples of roles and functions:

*Facilitator or leader* (depending on context) - to clarify the goals of the meeting and to summarize the discussions and decisions; ensures that the meeting takes place, continues and the basic rules are respected.

*Secretary* - keep a record of the ideas discussed and decisions made and who does what.

*Time Manager* - to make sure that you discuss everything that you need in the time allotted for the meeting.

*Controller* - to ensure that work is completed by an agreed time, and to solve problems if they are not being performed.

*A process observer* is someone who monitors the process, not the content, and can bring problems to the attention of the team. In this role, it is important to be positive, not condemning.

*Editor* - bring all materials together, identify gaps or matches and ensure consistency in the final presentation.

6. *Make it clear.* When a decision is made, it should be explained in such a way that it is absolutely clear to everyone that it was decided, including the time frame.

7. *Keep good notes.* Always summarize the discussions and document the decisions and publish them (for example in WhatsApp chat) so you can always get back to them. This includes lists of those who agreed what to do.

8. *Stick to the plan.* If you agreed to do something as part of the plan, do it. Your group relies on you to do what you agreed to do, and exactly in this way, not in the way you would like. If you think the plan should be reviewed, discuss it.

9. *Keep track of progress and keep up to date.* Discuss progress together regarding your schedule and deadlines. Make sure you meet deadlines personally so you do not let your group down.

### Co-writing a document / report

Joint writing is one of the most difficult parts of group work. There are many ways to do this, and your group must decide how to separate the work of writing, comparing, editing, and finalizing your work. Writing in a group (six people crowd around the keyboard) is a recipe for conflict and lack of progress. The other extreme - when one person assumes all responsibility and ultimately does most of the work - is also unproductive and contributes to conflict.

Three approaches are possible when working on a common document:

1 - One person writes the most part - this means that a narrow circle of ideas is used, and the rest of the team does not learn (and will not learn) to write reports and documents.

2 - Each person writes one section - then it is difficult to make a single consistent report, and you will not know about the rest, except for your own section.

3- Co-writing. This is the most productive way to solve group problems and provides the greatest benefit from collaboration. For example: in each section, there is a writer and at least one reviewer, and each team member is the author of a section and a reviewer of another one.

All team members before finalization by the editor must review the final product. Alternatively, you can have one author with others, editors, add and review, and someone tidies the finished report.

Try to divide the writing of source documents into tasks and solve them individually or in pairs. After the first draft of the sections are written, send out all the components and read them. You will probably need to come together to discuss how to combine them so that they fit together. Any participants who were not involved in preparing the drafts can do part of this work. Then edit, improve and polish the draft. It's convenient to collaborate on documents in Google documents.

When preparing a report / final document, regularly check the following:

- Is the purpose of the project clear from the report?
- Are the conclusions or recommendations clear?
- Do conclusions follow from the main part of the report?
- Do sections fit well?
- Does the report achieve goals (and evaluation criteria)?
- Are the necessary components sufficiently covered?

Whatever method you use, all group members must agree on the process and how they are going to maximize the collaborative approach to writing the final document.

### **Monitoring team performance and coping**

Below is a checklist that includes a list of common problems that arise in a group work. Use it regularly to identify problems before they get out of hand. If serious problems and tensions arise, use it to determine where something might go wrong. First answer each question about yourself, and then give answer to this question about the group as a whole. Then gather a group and discuss where, in your opinion, problems may arise, and think about how you can overcome these problems.

Each participant must complete this checklist. You should do this exercise regularly to track and improve your team's performance.

1. Answer each question regarding your teamwork.
2. Answer each question regarding the rest of the team.
3. Get together with your entire team and discuss where, in your opinion, any problems arise.
4. Discuss what you are going to do to overcome these problems.

### **Checklist for self-assessment of team effectiveness.**

You	Me personally	Group as a whole	Comments
Effectively clarify your tasks and tasks at each stage?			
Evaluate the progress of work?			
We clarify and document everything that the group decided?			
We clarify who will do what and how?			
We clarify by what date each task should be done?			
Setting meeting management rules?			
Adhere to agreed rules?			
Listening to each other?			
Allow some team members to dominate?			
Allow some team members to refuse / withdraw?			
We sacrifice personal desires for the success of the team?			
Recognize the feelings of other team members?			
Making equal contributions to team progress?			
Adhere to agreed rules for writing and naming files?			

### **Points and Grade**

Group tasks and assignments mean that grades are given to the whole group based on the results of the work of the whole group. Everyone should be interested in ensuring the effective contribution of all members of the group and ensuring the high quality of the assignment. Sometimes, to assess the relative contribution of each to the group process, a form of peer-to-peer or peer review and a team assessment form will be used. This can be used to moderate assignment grades, or simply as a way to give feedback on your work in a group. The following are examples of student assessment criteria for team training.

№	Student assessment criteria in practical classes
1	<i>Preparation for classes:</i> He studies information focused on the case and problematic issues, uses various sources, and supports the statements with relevant links.
2	<i>Group skills and professional attitude:</i> Demonstrates excellent attendance, reliability, responsibility Takes the initiative, takes an active part in the discussion, helps the teammates, willingly takes on tasks
3	<i>Communication skills:</i> Actively listens, shows emotions according to the situation, is susceptible to non-verbal and emotional signals, shows respect and correctness in relation to others, helps to resolve misunderstandings and conflicts
4	<i>Feedback Skills:</i> Demonstrates a high level of introspection, critically evaluates oneself and colleagues, provides constructive and objective feedback in a friendly manner, accepts feedback without opposition
5	<i>Skills of critical thinking and effective learning:</i> Effectively participates in generating hypotheses and formulating problematic questions, gives relevant examples from life, skillfully applies knowledge to the problem / case under consideration, critically evaluates information, draws conclusions, explains and substantiates statements, draws diagrams and drawings, demonstrates a constant interest in the material being studied
6	<i>Theoretical knowledge and skills on the topic of the lesson:</i> All key aspects are presented logically; accuracy, relevance of answers to the questions posed without redundancy; integration of theoretical issues; Use of relevant examples proper use of professional terminology

#### 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.
10. <https://www.msmanuals.com/professional/clinical-pharmacology>