INTERNATIONAL HIGHER SCHOOL OF MEDICINE

Special Clinical Disciplines Department

SYLLABUS
Medical genetics

2026-2027 academic year
for students of medical faculty
4th course VIIth semester, 1 – 40 groups of the Central Campus, 1-15 groups of the Issyk-Kul Campus
2 credits (60 h, including auditoria-36 h, individual work of student-24 h)

Lecturers: **Abdullaeva Nadira (Issyk-Kul campus)**
0703518454
Jusupova Asel Talgatbekovna (Central campus)
0550228845

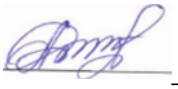
Lectures are held online via Zoom

Practical Classes **Abdullaeva Nadira (groups 22-26)**
0703518454
Sadyrbekova Shirin Zhenishbekovna (groups 27-32)
0550550717
Sayakbaeva Albina Kadyrbekovna (groups 1-9)
0 708 006 019
Jusupova Asel Talgatbekovna (groups 15-21)
0706282200
Kerimkulova Asel Kenzhebekovna (groups 33-40)
0551820729
Mataeva Gulzat Tursunbekovna (groups 10-14)
0559031771

Location:

1. Abdullaeva Nadira- Vedanta Plus, room 300
2. Jusupova Asel Talgatbekovna- Vedanta Plus, room 307
3. Kerimculova Asel Kenzhebekovna- Vedanta Plus, room 304
4. Mataeva Gulzat Tursunbekovna-Vedanta Plus, room 303
5. Sadyrbekova Shirin Zhenishbekovna- Virtual Clinic, room 110
6. Sayakbaeva Albina Kadyrbekovna- National Oncology Centre, room 005

The Syllabus is considered
at the meeting of the SKD department
Protocol №_1____dated_28_August ____2026

Head of the department__  ____ D.A.Maktybaeva

Course Objective: The main goal of the course consists of mastering knowledge of the main clinical forms of hereditary diseases, as well as the principles of clinical diagnosis and semiotics of hereditary diseases, skills and abilities in the treatment and prevention of genetic diseases

As a result of studying the discipline, the student must:

Know:

-Features of clinical manifestations and general principles of clinical diagnosis of hereditary diseases.

-General questions of etiology, pathogenesis, clinical features of hereditary diseases.

-Indications for the use of special examination methods.

-Methods of prevention and treatment of genetic disorders, principles of pathogenetic, symptomatic, and gene therapy.

-Stages and content of medical genetic counseling; indications for referring a patient for medical genetic counseling

Skill:

-Collect anamnestic data and genealogical information, create a pedigree, present it graphically and analyze the traits of inheritance of diseases running within a family.

-Select the patients to conduct cytogenetic, special biochemical and molecular genetic studies.

-Refer patients for medical and genetic counseling, provide the necessary documentation for medical and genetic consultation, help the family make decisions on family planning after medical and genetic consultation.

- Carry out preventive measures aimed at preventing hereditary and congenital diseases, reducing the incidence of widespread diseases of a multifactorial nature.

Attitude:

-Skills in analysis, evaluation of effectiveness and optimization of pathogenetically based methods of treatment, rehabilitation and prevention of the most common genetic diseases in various age groups of patients.

-Skills in substantiating and formulating a diagnosis based on the results of clinical, laboratory and functional examinations of adults and children

-The ability to substantiate one's own position on scientific and medical research issues based on the principles of evidence-based medicine

Pre-requisites. Macro and microanatomy, physiology, pathological anatomy, pathological physiology, internal diseases propedeutics, nervous system(clinical module), neurology with basics of neurosurgery.

Post-requisites. Pediatrics

THEMATIC PLAN OF LECTURES

№	Theme of lecture	Hours	Date
Unit No. 1: "Chromosomal diseases. Hereditary diseases affecting the muscular, pyramidal, extrapyramidal and cerebellar systems"			
1	Introduction to medical genetics. Classification of hereditary diseases. Methods of hereditary diseases diagnostics. Principles of genetic counselling	2	According to the schedule
2	Chromosomal diseases: Down, Patau, Edwards syndrome, Turner, Klinfelter, Prader Willi syndrome.	2	-
3	Hereditary muscular dystrophies	2	-
4	Disorders with damage of pyramidal and cerebellar systems (Hereditary spastic paraplegia, Friedreich's ataxia)	2	-
5	Lipid metabolism disorders (lipidoses) Disorders of carbohydrate metabolism (galactosemia) Amino acid metabolism disorders (phenylketonuria, histidinemia)	2	-
6	Mitochondrial encephalopathies.	2	-
7	Peroxisomal diseases	2	-
8	Clinical genetics of multifactorial diseases (diseases with predisposition).	2	-
	Total	16	

THEMATIC PLAN OF PRACTICAL CLASSES

№	Theme of practical class	Hours	Date
Unit No. 1: "Chromosomal diseases. Hereditary diseases affecting the muscular, pyramidal, extrapyramidal and cerebellar systems"			

1	Introduction to medical genetics. Classification of hereditary diseases. Methods hereditary diseases diagnostics. Chromosomal diseases: Down, Patau, Edwards syndromes.	2	According to the schedule
2	Prader Willi syndrome, Turner, Klinfelter. Discuss the referral criteria and multidisciplinary approach to management of Turner Syndrome, Prader Willi syndrome. Principles of genetic counselling. Describe the various modes of inheritance with examples.	2	
3	Hereditary muscular dystrophies (Duchenne muscular dystrophy, Beker, Emery-Dreifuss, Facioscapulohumeral muscular dystrophy, Limb-girdle muscular dystrophy). Spinal muscular atrophies (spinal muscular atrophy of Werdnig-Hoffman and Kugelberg-Welander)	2	-
4	Disorders with damage of pyramidal and cerebellar systems (Hereditary spastic paraplegia, Friedreich's ataxia) Degenerative diseases of CNS (hepatolenticular degeneration, Parkinson's disease)	2	-
5	Test 1	2	-
Unit No. 2: Disorders with lipid, carbohydrates, amino acids metabolism. Mitochondrial encephalopathies, and peroxisomal disorders			
6	Lipid metabolism disorders (lipidoses) Disorders of carbohydrate metabolism (galactosemia) Amino acid metabolism disorders (phenylketonuria, histidinemia)	2	-
7	Mitochondrial encephalopathies (MELAS)	2	-
8	Peroxisomal diseases (Zellweger syndrome, Refsum's disease)	2	-
9	Multifactorial disorders (Alzheimer's disease, Schizophrenia)	2	
10	Test 2	2	
	Total	20	

THEMATIC PLAN OF INDEPENDENT WORK OF STUDENTS

№	Theme of practical class	Hours	Date
Unit No. 1: "Chromosomal diseases. Hereditary diseases affecting the muscular, pyramidal, extrapyramidal and cerebellar systems"			
1	Make a pedigree on A4 paper. a) Indicate all symbols c) Give a brief description of each 2. Outline the purpose and objectives of genealogical analysis. 3. Make a report on the topic "Diagnostic value of the study of sex chromatin" 4. Draw the one for the various types of inheritance & give examples of diseases of each mode of inheritance	6	According to the schedule
2	a) To make a table on A4 paper with classification and description of each type of SMA b) Genetic data Spinal muscular atrophies (spinal muscular atrophy of Werdnig-Hoffman and Kugelberg-Welander) To make notes on clinical characteristics and laboratory data Criteria of diagnosis Differential diagnosis.	6	-
Unit No. 2: Disorders with lipid, carbohydrates, amino acids metabolism. Mitochondrial encephalopathies, and peroxisomal disorders			
3	1.Make a presentation on the topic: "State and prospects of genomic research in cardiology, pulmonology and nephrology" "General characteristics of a diseases with a hereditary predisposition (myocardial infarction, diabetes mellitus, leukemia) Classification. Etiology and pathogenesis. Clinical and genealogical evidence of hereditary predisposition" 1. Make a PPT on the topic: "Treatment and prevention of hereditary diseases.	12	-

	Total	24	

Recommended reading for the discipline:
Basic literature:

№	Авторы	Заглавие	Год издания	Издательство	Наличие в библиотеке МВШМ (количество)
1	Laura M. Gunder, Scott A. Martin	"Essentials of Medical Genetics for Health Professionals"	2012	Jones & Bartlett Learning	https://books.google.com/books?uid=111238161718902875581&as_coll=0&hl=ru&source=gbs_lp_bookshelf_list
2	Nicholas Wood	"Neurogenetics: A Guide for Clinicians"	2012	Published in the United States of America by Cambridge University Press, New York	https://books.google.com/books?uid=111238161718902875581&as_coll=0&hl=ru&source=gbs_lp_bookshelf_list
3	Robert M. Kliegman	Nelson texbook of pediatrics	2023	Elsevier	https://books.google.com/books?uid=111238161718902875581&as_coll=0&hl=ru&source=gbs_lp_bookshelf_list
4	G.P.Pal	"Medical genetics"	2009	AITBS Publishers of India	https://books.google.kg/books?id=7Qh-DwAAQBAJ&printsec=frontcover&source=gbs_ge_summary_r&cad=0#v=onepage&q&f=false

Additional literature

№	Авторы	Заглавие	Год издания	Издательство	Наличие в библиотеке МВШМ (количество)
1	Maurice Victor, Allan H.Ropper	"Manual of neurology"	2011	McGraw-Hill	https://books.google.com/books?uid=111238161718902875581&as_coll=0&hl=ru&source=gbs_lp_bookshelf_list
2	Stephen N.Adler, Mildred Lam	"A pocket manual of differential diagnosis"	2009	Lippincott Williams& Wilkins	https://books.google.kg/books?id=yIysxGug384C&printsec=frontcover&source=gbs_ge_summary_r&cad=0#v=onepage&q&f=false

Grading policy and procedures for all types of work
For the period of studying the discipline, the student gains points for the relevant parameters (per unit):
current score - 40 points
independent work - 20 points
control score (final assessment of knowledge per unit) - 40 points
Maximum score - 100 (40+20+40)
Grading system for student's achievements

Grading criteria per discipline

Maximum score	Intervals			
	«unsatisfactory»	«satisfactory»	«good»	«excellent»
Current control– 40	0-23	24-30	31-35	36-40
Interval description	the student has not mastered the required minimum of knowledge on the topic, did not answer questions regarding disciplines, studied previously	the student has the basic knowledge on the topic; has difficulty in answering independently, uses imprecise formulations; mistakes are made when student answers	the student knows the topic, is able to solve easy and more than average case study; is able to interpret laboratory and instrumental studies in a volume exceeding the mandatory minimum	the student knows the topic in full range
Individual work of student– 20	0-11	12-15	16-17	18-20
Interval description	incomplete (less than 50 %) an assignment has been done; many mistakes have been made	incomplete (no less than 50 %), but an assignment has been done, a meaningful mistake has been made; knows and understands the main points of topic, but answers are given with difficulties	incomplete (at least 70%), but the task is presented correctly; during the presentation, 1-2 insignificant errors were made, which he corrects after the teacher's remark; gives correct formulations, precise definitions, concepts of terms; can justify his answer, give the necessary examples; correctly answers additional questions	The student receives the maximum score for each type of assignment if: he presents the relevant topic in detail and with sufficient completeness; gives correct formulations, precise definitions, concepts of terms; correctly answers additional questions
Control work (module) – 40	0-23	24-30	31-35	36-40
Interval description	the student has not mastered the required minimum knowledge of the subject, is unable to answer questions even with additional questions from the examiner, or score is less than 60% during test	the student has the basic knowledge of the discipline; has difficulty in answering independently, uses imprecise formulations; When answers, errors are made	the student has knowledge of the discipline almost to the full extent of the program (does not always highlight the most significant, but at the same time does not make serious mistakes in answers; knows how to solve easy and more than average cases; knows how to interpret laboratory and instrumental studies in a volume exceeding the mandatory minimum	the student has knowledge of the subject in the full scope of the curriculum. Able to analyze, compare, classify, generalize, specify and systematize the studied material, highlight the main points in it: establish cause-and-effect relationships; clearly forms answers, interpret results of analyzes and other studies; is well acquainted with the basic literature and research methods to the extent necessary for practical activities; connects the

				theoretical aspects of the subject with practical tasks, has knowledge of the basic principles of the discipline. When solving test, answers 90% or more questions correctly

In case of class being missed, the overall score for the current subject is reduced by the following way: by 2 points if 25% of classes are missed, by 5 points, if from 25%-50%, and if 50% or more classes- by 10 points

Conduct Policy: (lateness, absence, behavior in the auditorium, late submission of work).
- Punctuality and completion of tasks.
- Mandatory attendance of classes.
- Attending class in a clean medical uniform.
- Eliminating conversations on a cell phone in the classroom.
- Active participation in the learning process.
- Doing homework on time.
- Academic detention at the time specified by the teacher.

For violations of the Conduct Policy, the total points for discipline might be reduced to 1-10 points.

Academic Ethics Policy.
- Be tolerant, respect the opinions of others.
- Formulate objections in the correct form.
- Constructively support feedback in all classes.
- Plagiarism and other forms of dishonest work are unacceptable. Plagiarism includes the following: the absence of references when using printed and electronic materials, quotes, thoughts and works of other authors or students.
- Prompting and cheating during tests, exams, classes is unacceptable as well as passing an exam for another student, unauthorized copying of materials.

For violations of the Academic Ethics Policy, the total points for the discipline may be reduced to 1-10 points.

Learning objectives
Key questions covered in Practical class 1: Introduction to medical genetics. Classification of hereditary diseases. Methods are used in genetics.
2 hrs.
1. Basic concepts in medical genetics.
2. Variability of hereditary traits
3. The role of heredity.
4. Classification of hereditary diseases.

Recommended literature for the class:
Basic literature:
1. Lecture course on Medical Genetics
2. "Essentials of Medical Genetics for Health Professionals", Laura M. Gunder, Scott A. Martin, 2012; P.1-17, 21-29
3. "Neurogenetics: A Guide for Clinicians", Nicholas Wood, 2012; P. 1-6
4. "Nelson texbook of pediatrics", Robert M. Kliegman, 2023; P. 177-192

Learning objectives
Key questions covered in Practical class 2: Chromosomal diseases. Types. Risk factors. Clinical manifestation; Diagnosis; Management.
2 hrs.
1. Prader Willi syndrome, Turner, Klinfelter.
2. Discuss the referral criteria and multidisciplinary approach to management of Turner Syndrome, Prader Willi syndrome.
3. Principles of genetic counselling.
4. Describe the various modes of inheritance with examples.

Recommended literature for the class:
Basic literature:
5. Lecture course on Medical Genetics

6. “Essentials of Medical Genetics for Health Professionals”, Laura M. Gunder, Scott A. Martin, 2012; P.1-17, 21-29
7. “Neurogenetics: A Guide for Clinicians”, Nicholas Wood, 2012; P. 1-6
8. “Nelson texbook of pediatrics”, Robert M. Kliegman, 2023; P. 177-192

Learning objectives

Key questions covered in Practical class 3: Hereditary muscular dystrophies. Spinal muscular atrophies.
2 hrs.

1. Classification of dystrophies. Clinical manifestation. Diagnosis, treatment
2. Duchenne muscular dystrophy
3. Beker, Emery-Dreifuss
4. Facioscapulohumeral muscular dystrophy
5. Limb-girdle muscular dystrophy.
6. Spinal muscular atrophies: Werdnig-Hoffman and Kugelberg-Welander

Recommended literature for the class:

Basic literature:

1. Lecture course on Medical Genetics
2. “Neurogenetics: A Guide for Clinicians”, Nicholas Wood, 2012; P. 148-166
3. “Nelson texbook of pediatrics”, Robert M. Kliegman, 2023; P. 710-182

Learning objectives

Key questions covered in Practical class 4: Disorders with damage of pyramidal and cerebellar systems (Friedreich's ataxia). Degenerative diseases of CNS (hepatolenticular degeneration, Parkinson’s disease)

1. Friedreich's ataxia
2. Hepatolenticular degeneration
3. Parkinson’s disease
4. Aetiology, pathogenesis, clinical manifestation, up-to-date methods of diagnosis, treatment and prevention

Recommended literature for the class:

Basic literature:

1. Lecture course on Medical Genetics
2. “Essentials of Medical Genetics for Health Professionals”, Laura M. Gunder, Scott A. Martin, 2012; P.716-717, 21-29
3. “Neurogenetics: A Guide for Clinicians”, Nicholas Wood, 2012; P. 52-64, 64-83
4. “Nelson textbook of pediatrics”, Robert M. Kliegman, 2023; P. 177-192

Learning objectives

Key questions covered in Practical class 5: Test/Module
1.Calculation of individual rating for the first module
2 hours

1. Viva
2. MCQ-based test on google form

Learning objectives

Key questions covered in Practical class 6: Lipid metabolism disorders (lipidoses); Disorders of carbohydrate metabolism (galactosemia); Amino acid metabolism disorders (phenylketonuria, histidinemia)
2 hours.

1. Lipid metabolism disorders (lipidoses)
2. Disorders of carbohydrate metabolism (galactosemia)
3. Amino acid metabolism disorders (phenylketonuria, histidinemia)
4. Aetiology, pathogenesis, clinical manifestation, up-to-date methods of diagnosis, treatment and prevention

Recommended literature for the class:

Basic literature:

1. Lecture course on Medical Genetics
2. “Essentials of Medical Genetics for Health Professionals”, Laura M. Gunder, Scott A. Martin, 2012; P.201-223
3. “Nelson textbook of pediatrics”, Robert M. Kliegman, 2023; P.210-212
4. “Medical genetics”, G.P. Pal, 2009; P. 176-182

Learning objectives

Key questions covered in Practical class 7: Mitochondrial encephalopathies (MELAS)
2 hours

1. Genetics of mitochondrial disorders

2. Epidemiology of mitochondrial disorders
3. Clinical features of patients with defects of mitochondrial disorders
4. MELAS
5. LHON **Key questions covered in**

Recommended literature for the class:

Basic literature:

1. Lecture course on Medical Genetics
2. "Essentials of Medical Genetics for Health Professionals", Laura M. Gunder, Scott A. Martin, 2012; P.183-189, 190-205
3. "Nelson textbook of pediatrics", Robert M. Kliegman, 2023; P. 223-225
4. "Neurogenetics: A Guide for Clinicians", Nicholas Wood, 2012; P.188-200, 201-212

Learning objectives

Key questions covered in Practical class 8: Peroxisomal diseases (Zellweger syndrome, Refsum's disease)

2 hours

1. Zellweger syndrome
2. Diagnostic testing
3. Treatment strategies
4. Refsum's disease

Recommended literature for the class:

Basic literature:

1. Lecture course on Medical Genetics
2. "Nelson textbook of pediatrics",Robert M. Kliegman, 2023; P. 219-223

Learning objectives

Key questions covered in Practical class 9: Multifactorial disorders (Alzheimer's disease, Schizophrenia)

1. Alzheimer's disease.
2. Its risk factors, pathogenesis, clinical manifestation, diagnosis, treatment, and prevention strategies
3. Schizophrenia as a multifactorial chronic psychotic disorder

Recommended literature for the class:

Basic literature:

1. Lecture course on Medical Genetics
2. "Essentials of Medical Genetics for Health Professionals", Laura M. Gunder, Scott A. Martin, 2012; P.45-53, 53-60
3. "Neurogenetics: A Guide for Clinicians", Nicholas Wood, 2012; P.17-32

Learning objectives

Practical class 10: final class. Test/Module 2. Calculation of individual rating along with overall score

1. Viva
2. MCQ-based test

Methodological instructions for independent work:

Unit No. 1: "Chromosomal diseases. Hereditary diseases affecting the muscular, pyramidal, extrapyramidal and cerebellar systems"

Learning objectives

Individual work of student theme 1

6 hours

1. Make a pedigree on A4 paper.
2. a) Indicate all symbols
3. c) Give a brief description of each
4. Outline the purpose and objectives of genealogical analysis.
5. Make a report on the topic "Diagnostic value of the study of sex chromatin"
6. Draw the one for the various types of inheritance & give examples of diseases of each mode of inheritance

Recommended literature:

Basic literature:

1. Lecture course on Medical Genetics
2. "Essentials of Medical Genetics for Health Professionals", Laura M. Gunder, Scott A. Martin, 2012; P.1-17, 21-29
3. "Neurogenetics: A Guide for Clinicians", Nicholas Wood, 2012; P. 1-6
4. "Nelson texbook of pediatrics", Robert M. Kliegman, 2023; P. 177-192

Learning objectives
Individual work of student theme 2
6 hours
1. Make a table on A4 paper with classification and description of each type of SMA
2. Genetic data
3. Spinal muscular atrophies (spinal muscular atrophy of Werdnig-Hoffman and Kugelberg-Welander)
4. Make notes on clinical characteristics and laboratory data
5. Criteria of diagnosis
6. Differential diagnosis.

Recommended literature:
Basic literature:

1. Lecture course on Medical Genetics
2. "Neurogenetics: A Guide for Clinicians", Nicholas Wood, 2012; P. 148-166
3. "Nelson textbook of pediatrics", Robert M. Kliegman, 2023; P. 710-182

Unit No. 2: Disorders with lipid, carbohydrates, amino acids metabolism. Mitochondrial encephalopathies, and peroxisomal disorders
Learning objectives
Individual work of student theme 2
12 hours
1.Make a presentation on the topic: "State and prospects of genomic research in cardiology, pulmonology and nephrology"
2. "General characteristics of diseases with a hereditary predisposition (myocardial infarction, diabetes mellitus, leukemia)
3. Classification. Etiology and pathogenesis.
4. Clinical and genealogical evidence of hereditary predisposition"
5. Make a PPT on the topic: "Treatment and prevention of hereditary diseases.
Recommended literature:
Basic literature:
1. Lecture course on Medical Genetics
2. "Essentials of Medical Genetics for Health Professionals", Laura M. Gunder, Scott A. Martin, 2012; P.1-17, 21-29, 45-53, 53-60
3. "Neurogenetics: A Guide for Clinicians", Nicholas Wood, 2012; P. 1-6, 17-32
4. "Nelson textbook of pediatrics", Robert M. Kliegman, 2023; P. 177-192