МЕДИЦИНСКИ УНИВЕРСИТЕТ – СОФИЯ
Medical University - Sofia
МЕДИЦИНСКИ ФАКУЛТЕТ
Faculty of Medicine – Dean's Office

MEDICAL GENETICS CURRICULUM

Approved by:
Corresp. Member of BAS Prof. I. Mitov, MD, PhD, DMSc
Dean of the Medical Faculty at
the Medical University- Sofia

The curriculum has been adopted at the meeting of the Faculty Council .....

Academic discipline:	**MEDICAL GENETICS**
Degree programme:	**MEDICINE**
Educational-qualification degree:	**MASTER**
Type of discipline in accordance with the Uniform State Requirements/ Curriculum:	**COMPULSORY**
Duration of the course of study:	**WINTER SEMESTER**
Exam:	**YES**
Semester in which the exam is held:	**V**
Total number of hours of auditorium workload:	**60 academic hours**
Lectures	**30 academic hours**
Seminars	**30 academic hours**
Department:	**MEDICAL GENETICS**
Training base:	**MEDICAL BIOLOGICAL COMPLEX**

CURRICULUM

DISCIPLINE	Exams in semester	ACADEMIC HOURS			ALLOCATION PER SEMESTER					
		Total	Lectures	Seminars	I	II	III	IV	V	VI
MEDICAL GENETICS	V	60	30	30	--	--	--	--	2/2	--

1. Annotation of the academic discipline

Medical genetics is one of the fastest growing fields in medicine. It gives an in-depth knowledge of the genetic mechanisms underlying diseases in all fields of medicine - pediatrics, hematology, oncology, neurology, nephrology, ophthalmology, endocrinology, cardiology, obstetrics and gynecology, etc.

The aim of the theoretical and practical training in Medical Genetics is to build an up-to-date knowledge of human genetic pathology - molecular basis of hereditary structures; etiology, pathogenesis, inheritance, classification, clinical and genetic heterogeniety of hereditary diseases; to familiriaze the students with the current trends in the clinical practice for diagnosis, therapy and prevention of monogenic and polygenic pathology, chromosomal diseases and hereditary predispositions. Focal points of the student training in medical genetics are the acquisition of skills for genetic diagnosis - new diagnostic approaches, recognizing the characteristic syndromal constellations and the construction of differential diagnosis; acquiring working knowledge of the principles of current approaches for diagnosis, prevention and treatment; developing the ability to calculate risks in families with genetic condition; familiarizing the students with the basic legal and ethical standards in diagnosis and prevention of genetic pathology; acquiring working knowledge of the principles, organization and tasks of medical genetic counseling, the approaches and indications for prenatal and pre-implantation diagnostics, the organization and nature of mass and selective genetic screening programs.

2. Main objectives of the study programme

Acquiring basic theoretical clinical knowledge of the etiology, pathogenesis, inheritance, classification, clinical and genetic heterogeneity of genetic diseases; application in clinical practice of the current trends in diagnosis, therapy and prevention of monogenic and polygenic pathology, chromosomal diseases, genetic predispositions and malignancies. Acquiring basic practical skills for making a genetic diagnosis of monogenic, chromosomal, malignant diseases and genetic predispositions; for risk assessment in families with genetic disease; for the treatment and prevention options of genetic diseases.

3. Expected outcomes

3.1. Amount of acquired theoretical knowledge: understanding of the basic clinical and diagnostic criteria for common monogenic and chromosomal diseases, knowledge of the set of clinical and paraclinical tests used in diagnosis of genetic diseases, and the steps of the diagnostic process; knowledge of the principles and indications for cytogenetic, molecular-genetic and biochemical diagnostic tests; understanding of the benefits and limitations of predictive testing for genetic diseases; knowledge of the basic principles of genetic counseling; knowledge of the indications and main aspects of prenatal diagnosis; knowledge of the current screening programs for genetic diseases; knowledge of the major genetic derminants of adverse drug reactions drug and ineffective pharmacotherapy; understanding the role of germline and somatic mutations for oncological diseases and their place in the diagnosis, prognosis, treatment and prevention of malignancies; knowledge of the role of genetic testing in developing more effective approaches to maintaining health and personalized treatment;

3.2. Acquired practical skills: building pedigrees; recognizing type of inheritance patterns; recognizing the clinical signs suggestive of genetic disease in the family; identification of patients with predisposition to common diseases; ability to recognize syndromal constellations, congenital anomalies and their possible etiology; determination of genetic risk in affected families with monogenic, chromosomal or multifactorial disease; understanding and interpretation of the results of cytogenetic, molecular-genetic and biochemical diagnostic tests; working knowledge of the algorithm of the current genetic screening programs;

4. Thematic unit plan for lectures and seminars

4.1.Lectures - 30 academic hours (15 lectures in 2 academic hours)

1. Puropose, subject, tasks of Medical Genetics. Organziation of the Human genome
2. Congenital anaemias. Coagulation disorders. Cystic fibrosis
3. Metabolic disorders
4. Trinucleotide repeat disorders. Connectove tissue disorders
5. Neuromuscular disorders. Mitochondrial diseases
6. Polygenic diseases
7. Structure and functioin of the chromosomes. Diagnostic methods for chromosomal aberrations.
8. Chromosomal diseases. Microdeletion syndromes
9. Pharmacogenetics

10. Cancer genetics
11. Reproductive genetics. Dysmorphology and teratology
12. Genetics of hearing loss. Immunodeficiency syndromes.
13. Prenatal, preconception and preimplantational diagnostics.Genetic screening
14. Genetic diseases treatment .
15. Genetic methods. Medical genetic counseling

4.2.Seminars - 30 academic hours (15 seminars in 2 academic hours)

1. Types of inheritance
2. Thalassemias and cystic fibrosis
3. Metabolic diseases
4. Trinucleotide repeat disorders
5. Herditary connective tissue disorders
6. Neuromuscular disorders. bleeding disorders
7. Mitochondrial diseases. colloquium i
8. Normal human karyotype. chromosomal aberrations
9. Chromosomal diseases
10. Genomic imprinting. microdeletion syndromes
11. Oncogenetics. targeted therapy
12. Dysmorphology.teratology
13. Reproductive genetics
14. Prenatal diagnostics
15. Medical genetic counseling. Colloquium II

5. Supplementary teaching materials: workbook, photos, clinical cases, patients' test results, videos etc.

6. Assessment of acquired knowledge

6.1. Ongoing assessment

- Oral examination at the beginning of seminars
- Oral presentations
- Colloquiums consisting of a test and a clinical case

6.2 Semester exam

- Test
- Practical examination consisting of two clinical cases
- Theoretical exam – written summary of the topics, that are kept at the archive, and oral presentation of the topics before an examination committee