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:	**PHARMACY**
Educational-qualification degree:	**MASTER**
Type of discipline in accordance with the Uniform State Requirements/ Curriculum:	**COMPULSORY**
Duration of the course of study:	**IV**
Exam:	**YES**
Semester in which the exam is held:	**VIII**
Total number of hours of auditorium workload:	**30 academic hours**
Lectures	**15 academic hours**
Seminars	**15 academic hours**
Department:	**MEDICAL GENETICS**
Training base:	**MEDICAL BIOLOGICAL COMPLEX**

CURRICULUM- DEGREE PROGRAMME "PHARMACY"

Academic discipline	Exam/ semester	Academic hours Total	Lectures	Seminars	Часове по години								
			semester		*I year*		*II year*		*III year*		*IV year*		*V year*
					I	II	III	IV	V	VI	VII	VIII	IX
MEDICAL GENETICS	**VIII**	**30**	**15**	**15**	**-**	**-**	**-**	**-**	**-**	**-**	**-**	**1/1**	**-**

1. Annotation of the academic discipline

Medical genetics is one of the fastest growing fields in medicine. The aim of the theoretical and practical training in Medical genetics is for students to acquire an up-to-date knowledge of human hereditary pathology. The course covers the basic principles of human genetic pathology - etiology, pathogenesis, inheritance, modern approaches to diagnostics, prevention and treatment. A major focal point of the lecture and practical course are congenital anomalies and the role of teratogenic factors in their etiology. The course also emphasizes on the problems of pharmacogenetics and pharmacogenomics - genetic determinants of therapeutic response and adverse drug reactions. Students acquire working knowledge of genetic testing's place in personalized and precision therapy of oncological and other common diseases; of current approaches to pharmacotherapy for rare genetic diseases with emphasis on orphan drugs and options and conditions for gene therapy. The course also covers the basic aspects of genetic prevention - medical genetic counseling, prenatal diagnostics, and genetic screening programs.

2. Main objectives of the study programme

Acquiring basic theoretical clinical knowledge of the etiology, pathogenesis, inheritance, clinical presentation and current approaches in the treatment of common hereditary diseases with monogenic and chromosomal etiology; acquiring in-depth knowledge of the role of genetic factors for oncogenesis; acquiring working knowledge of the application of genetic tests that can predict adverse reaction to commonly used medications and that are part of cancer treatment choice algorithms; in-depth knowledge of the principles of the most widespread molecular-genetic and cytogenetic methods.

3. Expected outcomes

3.1. Amount of acquired theoretical knowledge: an understanding of the basic aspects of hereditary structures and the flow of genetic information; knowledge of the characteristics of the main types of genetic diseases - monogenic, chromosomal and malignant; an understanding of the genetic mechanisms for cell cycle regulation; basic knowledge of the clinical presentation of the most common monogenic and chromosomal diseases and current treatment options; understanding of the genetic factors of therapeutic response and their role in determining dosage for some commonly prescribed medications; knowledge of the role of genetic testing in cancer treatment; an in-depth knowledge of teratogenicity determinants of pharmacotherapy during pregnancy.

3.2. Acquired practical skills: being able to determine type of inheritance; recognizing the major signs suggestive of genetic disease in the family; working knowledge of the principles, advantages and limitations of cytogenetic, molecular-genetic and biochemical diagnostic tests; working knowledge of the algorithm of current screening programs; being able to suggest dosage modification for some commonly prescribed medications based on patient's genetic status; working knowledge of the indications for genetic testing before onset of treatment; working knowledge of the indications for genetic testing for cancer treatment; being able to recognize the clinical symptoms associated with prenatal exposure to some common pharmacological agents.

4. Thematic unit plan for lectures and seminars

4.1.Lectures – 15 academic hours (6 lectures in 2academic hours, 1 lecture in 3 academic hours)

1. Subject, tasks and aspects of Medical genetics. Heredity – structures, composition and organization of DNA. Organization of hereditary information.
2. Mutagenesis. Medicines as mutagens. Types of mutations. Classification of genetic diseases.
3. Birth defects. Genetic and non-geentic causes. Teratogenic effect of drugs.
4. Pharmacogenetic defects (PhGDs).General characteristics. Classification. Monogenic PhGDs.
5. Pharmacogenetic defects. Liver cytochrome P450 monooxygenase polymorphisms. PhGDs in genetic diseases.
6. Genetic regulation of cell proliferation, cell differentiation and apoptosis. Carcinogenesis.
7. Treatment of genetic diseases. Conventional therapy and gene therapy. Current trends.

4.2.Seminars -15 academic hours (6 seminars in 2 academic hours, 1 seminar in 3 academic hours)

1. Structure of the human genome. Humangenome variability. Genetic methods. Types of ihteritance
2. Chromosomal mutations. Clastogenic effect of drugs
3. Precision medicine and pharmacogenetic defects (PhGDs)
   Pprecision therapy based on *CYP2D6, CYP2C9, CYP2C19,* and *HLA* genetic variants
4. Monogenic diseases and orphan drugs.
5. Oncogenetics and targeted therapy
6. Dysmorphology and teratology

5. Supplementary teaching materials: workbook, video materials

6. Assessment of acquired knowledge

6.1. Ongoing assessment

- Oral examination at the beginning of every seminar
- Oral presentations

6.2 Semester exam

- Test
- Practical examination consisting of two clinical cases/tasks