Катедра по медицинска генетика, Медицински Факултет, Медицински Университет-София, ул. Здраве 2, 1431 София, medgenetics@medfac.mu-sofia.bg

Department of Medical Genetics, Medical Faculty, Medical University of Sofia, 2 Zdrave str, medgenetics@medfac.mu-sofia.bg

MEDICAL GENETICS

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.

Aims

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.

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.