UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Genetic Disease

2627-2-I0301D007-I0301D027M

Aims

The student must be able to:

Describe the mechanisms underlying genetic diseases.
Describe atypical inheritance mechanisms.
Describe diseases caused by imprinting defects, dynamic mutations, mitochondrial and multifactorial disorders.
Describe examples of cancer predisposition.

Contents

By the end of the course, the students will have acquired the general concepts and specific knowledge of ethiopathogenesis of genetic diseases

Detailed program

Classification and incidence of genetic diseases.
Gene variants: origin, classification, and pathogenic effect.
Modes of transmission of genetic diseases: autosomal dominant and recessive, pedigree analysis, penetrance, expressivity, new mutations, mosaicism.
Concepts of clinical heterogeneity, locus heterogeneity, genotype-phenotype correlation.
Monogenic diseases with Mendelian inheritance and their effects on the phenotype; gain-of-function and loss-of-function mutations; examples of diseases. Sex-linked diseases.
Non-Mendelian inheritance: 1) Diseases caused by triplet repeat expansions (Huntington's disease and Fragile X syndrome); the concept of genetic anticipation; 2) Epigenetics and imprinting-related diseases: Angelman

syndrome and Prader-Willi syndrome; 3) Mitochondrial diseases. Numerical and structural chromosomal abnormalities.
Overview of multifactorial diseases: the role of DNA polymorphisms and the concept of genetic susceptibility.
Cancer predisposition syndromes: oncogenes and tumor suppressor genes, pediatric cancer predisposition syndromes, predisposition to pediatric acute lymphoblastic leukemia.
Classification of genetic tests, role of genetic counseling.

Prerequisites

basic concepts of mendelian genetics

Teaching form

Four two-hours frontal lessons in attendance

Textbook and teaching resource

Teacher's slides

Thompson and Thompson, Genetics in medicine, 8 ed. Elsevier
Strachan & Reid, Human Molecular Genetics, 4 Ed. Garland Science

Semester

First semester

Assessment method

- Written exam: 10 closed-ended questions (True/False, Matching, Multiple Choice, etc.)
  The assessment will evaluate the knowledge acquired during the lectures.

Office hours

On request by e-mail

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION | GENDER EQUALITY