UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Genetic Pathology

2425-2-I0303D007-I0303D032M

Aims

The student will be able to:
Describe the mechanisms underlying genetic diseases; Describe the atypical mechanisms of inheritance; Describe diseases caused by imprinting defects, dynamic, mitochondrial and multifactorial mutations; Describe examples of predisposition to tumors

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-based pathologies
Gene variants: origin, classification and pathogenic effect
Modes of transmission of genetic diseases: autosomal dominant and recessive, tree analysis, penetrance, expressivity, neomutation, mosaicism
Concepts of clinical heterogeneity, locus, genotype-phenotype correlation.
Monogenic diseases with Mendelian inheritance and effects on the phenotype; gain- and loss-of-function mutations; examples of pathologies. Sex-related diseases.
Non-Mendelian inheritance: 1) Nucleotide triplet expansion diseases (Huntington's chorea and Fragile X syndrome); the concept of genetic anticipation; 2) Epigenetics and diseases related to imprinting: Angelman and Prader-Willi syndrome; 3) mitochondrial diseases. Numerical and structural chromosomal anomalies.
Notes on multifactorial diseases: the role of DNA polymorphisms and the concept of genetic susceptibility.

Cancer predisposition syndromes: oncogenes and suppressor genes, Pediatric tumor predisposition syndromes, predisposition to pediatric acute lymphoblastic leukemia.
Classification of genetic tests, role of genetic counseling

Prerequisites

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Teaching form

4 frontal lessons of 2 hours carried out in attendance

Textbook and teaching resource

Slides provided by the teacher
Thompson and Thompson, Genetics in Medicine, 8th ed. Elsevier
Strachan and Reid, Human Molecular Genetics, 4th Ed. Garland Science

Semester

First semester

Assessment method

Monza
Closed-answer test (multiple choice) of Genetic Pathology to check preparation on the exam programme.

Bergamo
Closed-answer test (multiple choice) and open-ended questions on Genetic Pathology to evaluate preparation on the teaching program and the ability to organize knowledge in a short discussion.

Office hours

On request by e-mail

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION