UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Patologia Genetica

2425-2-H4101D255-H4101D029M

Aims

The student must be able to:

- Understand the mechanisms underlying genetic diseases.
- Understand atypical inheritance mechanisms.
- Have an overview of diseases caused by imprinting defects, dynamic mutations, mitochondrial and multifactorial disorders.
- Understand the genetics of cancer predisposition.
- Understand the genetics of diseases for precision medicine.
- Have an overview of DNA sequencing analysis.

Contents

By the end of the course, the student must have acquired essential knowledge about genetic diseases, as well as fundamental pathogenetic and pathophysiological mechanisms.

Detailed program

- Classification and incidence of genetic disorders.
- 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, the role of genetic counseling, examples of clinical cases with DNA sequencing simulation.
- Oncogenetics in precision medicine: risk stratification in pediatric acute lymphoblastic leukemia.

Prerequisites

basic concepts of molecular biology

Teaching form

Lectures will be delivered in Italian, with no remote teaching activities planned.
Clinical cases with DNA sequencing analysis simulation.

Textbook and teaching resource

- Teacher’s slides
- Thompson and Thompson, Genetics in Medicine
- Strachan & Reid, Human Molecular Genetics

Semester

Second semester

Assessment method

There are no midterm exams.

- Written exam: 10 closed-ended questions (True/False, Matching, Multiple Choice, etc.) (0 points for incorrect or incomplete answers).
  The assessment will evaluate the knowledge acquired during the lectures.
  The final grade will encompass all pathology modules.

Office hours

By appointment via email.

Sustainable Development Goals

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