



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Patologia Genetica

2425-2-H4101D255-H4101D029M

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#### 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

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