



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Patologia Genetica

2425-2-I0302D007-I0302D027M

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#### 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 student must have acquired essential knowledge about genetic diseases, as well as fundamental pathogenetic and pathophysiological mechanisms.

#### 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**

## **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 test (multiple choice test consist of 20 questions) to evaluate knowledges about course program

## **Office hours**

On request by e-mail

## **Sustainable Development Goals**

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

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