



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## COURSE SYLLABUS

### Medical Genetics

2526-1-H4103D157-H4103D15702

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#### Aims

The course aims to provide and deepen the biological tools and fundamental genetic bases necessary for understanding human genetic diseases, within the framework of genomic medicine.

#### Contents

The cellular and molecular bases of human genetics will be explored in detail, and the organization of genes and the human genome, its modifications, and the DNA repair mechanisms implemented by cells will be thoroughly analyzed.

Significant emphasis will also be placed on the epigenome, its modifications, and the complex mechanisms regulating gene expression.

To support and enhance overall understanding, technologies for the analysis and modification of nucleic acids, next-generation sequencing (NGS), and the bioinformatic approaches required to interpret the results obtained using these methodologies will also be examined.

#### Detailed program

##### Cytogenomics

- Banding techniques and advanced methods for the study of cytogenetic abnormalities and the genome
- Fluorescence in situ hybridization (FISH) and its developments
- Array comparative genomic hybridization (array CGH)
- Optical Genome Mapping

##### Mechanisms Underlying Chromosomal Aberrations

- Chromosomal syndromes and genomic disorders

- Segmental duplications
  - Microdeletion and microduplication syndromes
- Prenatal Diagnosis: Non-Invasive and Invasive Tests
- Non-invasive prenatal testing (NIPT)
  - Chorionic villus sampling
  - Amniocentesis
  - Mosaicism
- UPD and Its Implications in Imprinting Disorders
- Cancer Cytogenetics
- Molecular Diagnostic Techniques in Medical Genetics
- Simulation of exome analysis and interpretation of genetic variants
  - Precision medicine: genetics in support of clinical protocols
  - Cancer predisposition, with a focus on acute lymphoblastic leukemia
  - Medical genetics: genetic counseling and diagnostic testing
  - Non-Mendelian inherited diseases: trinucleotide repeat expansions
- Clinical Cases

## **Prerequisites**

See the information provided in the course syllabus

## **Teaching form**

See the information provided in the course syllabus

## **Textbook and teaching resource**

See the information provided in the course syllabus

## **Semester**

See the information provided in the course syllabus

## **Assessment method**

See the information provided in the course syllabus

## **Office hours**

See the information provided in the course syllabus

## **Sustainable Development Goals**

GOOD HEALTH AND WELL-BEING | GENDER EQUALITY | REDUCED INEQUALITIES

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