UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

# COURSE SYLLABUS

## Medical Genetics

**2324-4-H4101D262-H4101D077M**

### Aims

To introduce the students to appropriateness of genetic prescriptions

### Contents

Main tecniques of cytogenetics, molecular cytogenetics and molecular genetics

### Detailed program

• choosing between the different technologies for the diagnosis of genetic diseases; applications, advantages and limitations in comparison: cytogenetics; molecular cytogenetics (FISH and CGH array, SNP array); molecular genetics (PCR, digital and real-time PCR, allele-specific amplification, hybridization with allele-specific oligonucleotides, Sanger and NGS sequencing, MLPA)
• genetic counseling
• genetic tests: classification (diagnostic, presymptomatic, predictive, pharmacogenomics); appropriateness of requests for genetic testing and clinical utility; genetic testing for research; ethical and social aspects of genetic testing; system for genetic variant classification; ability to read and understand information of a genetic report.
• Genetic and Genomic Databases
• genomic diseases and complex diseases
• cancer predisposition syndromes
• invasive and Non invasive prenatal genetic testing
• clinical cases

## Prerequisites

Those foreseen by the didactic regulations of the CdS

## Teaching form

Lessons

## Textbook and teaching resource

Texbook: Tom Strachan, Andrew Read: Genetica molecolare umana
Slides
Guidelines and scientific articles provided by the teacher

## Semester

First semester

## Assessment method

See Syllabus of Laboratory Medicine

## Office hours

Appointemt on request by e-mail

## Sustainable Development Goals

GOOD HEALTH AND WELL-BEING