



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Genetica Medica

2425-4-H4101D262-H4101D077M

---

#### Aims

The module's goals are to help students comprehend the role that genetic tests play in modern diagnosis, the appropriate circumstances under which genetic tests should be ordered, and how to correctly evaluate the findings of genetic and genomic investigations in a clinical setting.

#### Contents

Main techniques 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 and labs cases

## **Prerequisites**

Basic science knowledge

## **Teaching form**

Twelve frontal teaching lessons—eleven of which begin with direct instruction and end with interactive teaching with clinical cases, exercises, and FAQs—with the goal of providing a clinical-practical re-elaboration of the concepts presented in the first part of the lesson. One lesson is devoted to interactive teaching on the interpretation of genetic reports.

## **Textbook and teaching resource**

Textbook: Tom Strachan, Andrew Read: Genetica molecolare umana

Slides

Guidelines and scientific articles provided by the teacher

## **Semester**

First semester

## **Assessment method**

Evaluation of knowledges and skills. knowledge gained and reasoning skills on case analyses based on the concepts learned will be assessed.

see syllabus "medicina di laboratorio"

## **Office hours**

Appointemt on request by e-mail

gaia.roversi@unimib.it

## **Sustainable Development Goals**

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

---