UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

# COURSE SYLLABUS

## Medical Genetics

**2425-6-H4102D083-H4102D181M**

### Aims

- Acquire knowledge in collecting a patient's family history and building a family pedigree
- Gain knowledge in utilizing the most common databases that deal with genetic syndromes and diseases
- Become acquainted with some of the most common diagnostic techniques (Karyotype, CGH-array, FISH)
- Become acquainted with the Next Generation Sequencing (NGS) workflow (from the samples' preparation to the analysis).

### Contents

The clerkship will allow students to achieve skills in collecting the clinical history of a patient referred for the suspicion of a genetic diseases, focusing on the importance of the family pedigree.
In addition, the student will have the chance to follow the preparation of most common diagnostic techniques and have the chance to deal with a Next Generation Sequencing analysis.

### Detailed program

- Collecting a family history and building a family pedigree of a patient with a suspected genetic condition
- Experience in utilizing common databases (OMIM, Orphanet, GeneReviews) to acquire information on genetic syndromes
- Follow the preparation of some of the most common diagnostic techniques (Karyotype, CGH-array, FISH)
- Follow an example of a NGS analysis

## Prerequisites

Fundamentals of molecular genetics and medical genetics

## Teaching form

- Seminars
- Problem based learning
- Simulation activities
- experience in the ward and in the laboratory

## Textbook and teaching resource

none

## Semester

first term

## Assessment method

frequency and practical skills assessment during the internship

## Office hours

write an email to
maria.iascone@gmail.com

## Sustainable Development Goals

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