



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Patologia Genetica

2122-2-H4101D255-H4101D029M

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#### Aims

The primary goal of the course is to provide the students with the necessary tools to understand the pathological mechanisms of genetic diseases

#### Contents

Genetic pathology and molecular pathology

#### Detailed program

Mechanisms of genetic diseases. Classification of human diseases. Monogenic diseases. Genotype -Phenotype correlation. Excpetions to mendelian inheritance: mitochondrial disease anddiseases of unstable repeat expansion. Epigenetic and imprinting defects. X inactivation mediated diseases. Complex diseases.

#### Prerequisites

propaedeutic courses

#### Teaching form

Lectures

Lessons will be provided in presence, subject to any ministerial changes following the COVID pandemic situation

### **Textbook and teaching resource**

material provided by teacher

### **Semester**

second semester second year

### **Assessment method**

multiple choice test. possible oral examination.

### **Office hours**

by appointment by mail request

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