



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## COURSE SYLLABUS

### Genetic Pathology

2021-2-H4101D255-H4101D029M

---

#### Aims

The course aims to provide the student with the necessary tools for understanding the pathological mechanisms of genetic diseases

#### Contents

Genetic and molecular genetic pathology

#### Detailed program

Classification of genetic diseases. Monogenic diseases. Genotype-phenotype correlations. Exceptions to Mendelian inheritance: triplet diseases and mitochondrial diseases. Epigenetics and diseases: imprinting and inactivation of X and pathological implications. Oligogenic and multifactorial inheritance.

#### Prerequisites

Basic knowledge of heredity, of the structure of chromosomes and genes and of the regulatory mechanisms carried out in the preparatory courses of the degree course in medicine and surgery

#### Teaching form

Due to the restrictions imposed by the Covid-19 pandemic, the lessons will be recorded on the Kaltura platform and made available on the site. In case of return to normality, the usual frontal lessons will be held.

## **Textbook and teaching resource**

Related bibliographic indications will be provided

## **Semester**

second semester II year

## **Assessment method**

Multiple choice questions for the assessment of knowledge. A short oral question may be held at the discretion of the teacher if really needed. Any possible changes will be defined according to the evolution of the Covid-19 pandemic

## **Office hours**

by appointment upon motivated request to be sent to the e-mail address of the teacher

---