



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## COURSE SYLLABUS

### Medical Genetics - 5

2526-1-I0102D001-I0102D003M-T5

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#### Aims

Students must understand genetic and chromosomal disorders, the concepts and modes of inheritance of hereditary traits, and the mechanisms of 'non-traditional' pathogenesis.

#### Contents

The course aims to provide knowledge of genetic and chromosomal disorders; the concepts and modes of inheritance of hereditary traits; the 'non-traditional' pathogenetic mechanisms; as well as the foundations for the qualitative and quantitative understanding of biological phenomena.

#### Detailed program

Introduction to Genetics. Overview of Mendel's laws. Genetics of blood groups, Rh and MN antigens, overview of reproductive physiology. The karyotype: historical background, techniques, and indications for prenatal and postnatal diagnosis. Numerical chromosomal abnormalities, structural chromosomal abnormalities and their clinical consequences on phenotype and reproduction. Sex determination: overview, disorders caused by numerical alterations of sex chromosomes; X chromosome inactivation. Different inheritance patterns of Mendelian monogenic diseases (pedigrees and examples of genetic disorders): autosomal dominant; autosomal recessive; X-linked recessive; X-linked dominant. Non-Mendelian inheritance: triplet repeat expansion disorders, imprinting, and related syndromes.

## **Prerequisites**

Basic knowledge of biology.

## **Teaching form**

In-person delivery method in Italian language

## **Textbook and teaching resource**

Slides by the lecturer Chieffi G., Dolfini S., Malcovati M., Pierantoni R., Poli M., TENCHINI M.L. Biology and Genetics (2013) Edises - 4th ed.

## **Semester**

First semester

## **Assessment method**

There are no in itinere assessments. Written exam: 11 questions with closed-ended answers (True/False, Matching, Multiple choice, etc.) (0 points for incorrect or incomplete answers). The knowledge acquired during the lectures will be assessed. The evaluation will be comprehensive across the different modules.

## **Office hours**

On appointment by e-mail request

## **Sustainable Development Goals**

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

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