



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## COURSE SYLLABUS

### Medical Genetics

2223-1-I0101D001-I0101D003M

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#### Aims

The student must achieve the knowledge of the histological and anatomical bases essential for the study and understanding of the human physiology and pathology. He must also know: the structure and function of cellular components and molecular mechanisms; the chromosomal disorders; the concepts and methods of transmission of hereditary characteristics and of pathogenic mechanisms "non-traditional"; the qualitative and quantitative knowledge of biological phenomena.

#### Contents

The course aims to describe the organization of the human body; to explain how are cells and tissues organized to form organs and systems; to underline the functional correlations of micro- and macroscopic anatomy. It also aims to transmit the knowledge of the structure and function of the various components of eukaryotic cells, the molecular mechanisms involved in cell replication, the molecular mechanisms involved in gene expression; the chromosomal disorders and transmission pattern in Mendelian monogenic diseases as well as the basis for the qualitative and quantitative knowledge of biological phenomena for a correct application of therapies.

#### Detailed program

MEDICAL GENETICS - Introduction to Genetics. Notes of Mendel's laws. Blood group Genetics, Rh and MN antigens, notes on the reproduction physiology. Karyotype: history, techniques, clinical indication for pre-and and postnatal diagnosis. Numerical and structural chromosomal abnormalities and clinical consequences on phenotype and reproduction. Notes on sex development, sex chromosomes aneuploidies and related syndromes; X-chromosome inactivation. Different types of monogenic Mendelian Inheritance (family trees and examples of genetic diseases): autosomal dominant; autosomal recessive; X-linked recessive; X-linked dominant. Non-

mendelian genetic diseases caused by expansion of triplets and imprinting: examples of syndromes.

## **Prerequisites**

## **Teaching form**

Face to face lectures

## **Textbook and teaching resource**

MEDICAL GENETICS - Chieffi G., Dolfini S., Malcovati M., Pierantoni R., Poli M., Tenchini M.L. Biologia e Genetica (2013) Edises - IV ed.

## **Semester**

1 Year - 1 Semester

## **Assessment method**

Written examination: multiple choice

## **Office hours**

On appointment

## **Sustainable Development Goals**

GOOD HEALTH AND WELL-BEING

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