UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Medical Genetics

2425-1-I0101D001-I0101D003M

Aims

Students are expected to acquire knowledge regarding the chromosomal and genetic pathologies, the fundamentals and the transmission models of the hereditary traits and of the "non traditional", pathogenic mechanisms.

Contents

The course aims to transmit knowledge about the chromosomal and genetic patologies; the fundamentals and the transmission fashion of the hereditary traits and of the pathogenic, "non traditional" mechanisms, as well as the basis for the quantitative and qualitative knowledge of the biological processes.

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 notes, techniques, indications for pre- and postnatal diagnosis
- Numerical chromosomal abnormalities, structural chromosomal abnormalities, and their clinical consequences on phenotype and reproduction
- Sex determination: overview, pathologies from numerical abnormalities of sex chromosomes; X chromosome inactivation
- Different types of inheritance of Mendelian monogenic diseases (pedigree charts and examples of genetic diseases): autosomal dominant; autosomal recessive; X-linked recessive; X-linked dominant
- Non-Mendelian inheritance: triplet expansion diseases, imprinting, and related syndromes

Prerequisites

basic knowledge of biology

Teaching form

Lectures in Italian, no remote teaching activities are planned

Textbook and teaching resource

Material provided by the teacher: slides.

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

Semester

First semester

Assessment method

No interim tests

- Written exam: 11 questions with closed answers (True/False, Matching, Multiple Choice, etc.) (0 points for incorrect or incomplete answers)
- Evaluation of the knowledge acquired during the lessons
- The evaluation will be comprehensive for the different modules

Office hours

on demand, by email

Sustainable Development Goals

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