UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Medical Genetics

2627-1-I0104D001-I0104D00103

Aims

Students must understand genetic and chromosomal disorders, the concepts and modes of inheritance of hereditary traits, and the mechanisms of 'non-traditional' pathogenesis.
A) Knowledge and Understanding
B) Applying Knowledge and Understanding
C) Making Judgements
D) Communication Skills
E) Learning Skills

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. (2013) Biologia e Genetica. (IV ed.) Edises, Napoli.

Semester

First semester

Assessment method

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

By appointment by e-mail request

Sustainable Development Goals

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