UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Genetica Medica

1920-1-I0102D001-I0102D003M

Aims

The student must 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

Contents

The module 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

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

Lectures

Textbook and teaching resource

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

Semester

First year, I semester

Assessment method

Written examination- 10 multiple choice - Oral on evaluation of teacher

Office hours

by appointment