



UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Molecular Genetics

2021-3-I0302D035-I0302D046M

Aims

Models of mendelian disorders and effect of mutation on cellular function. Approaches used to study Chromosomal syndromes, microdeletion syndromes and imprinting disorders. Karyotype analysis. Genetic variation: mutation and polymorphism. Tools of molecular karyotype analysis. Bioinformatics.

Contents

The primary goal of the course is to provide tools for the understanding of the laboratory techniques to approach hereditary diseases. Provide knowledge of human molecular genetics and cytogenetics.

Detailed program

Human Mendelian diseases: transmission and phenotypic complexity. The genes and their role in genetic diseases. Human diseases deriving from uniparental dysomy and related techniques. Unstable repeat expansion and the related technique, mutations in the mitochondrial genome. Chromosomal syndromes and microdeletions. Karyotype and its analysis. FISH, CGH, Fiber FISH and molecular analysis. New genetic techniques. Genomics and Proteomics.

Prerequisites

Having passed the compulsory courses of the degree

Teaching form

Lectures

Textbook and teaching resource

The Teachers will provide educational materials

Semester

First semester

Assessment method

Written test: multiple choice test and open questions

Oral Test: discussion of written test

Office hours

On appointment requested by mail
