UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Genetica Medica

2627-1-H4103D157-H4103D15702

Aims

The course aims to provide and deepen the biological tools and fundamental genetic bases necessary for understanding human genetic diseases, within the framework of genomic medicine.

Contents

The cellular and molecular bases of human genetics will be explored in detail, and the organization of genes and the human genome, its modifications, and the DNA repair mechanisms implemented by cells will be thoroughly analyzed.
Significant emphasis will also be placed on the epigenome, its modifications, and the complex mechanisms regulating gene expression.
To support and enhance overall understanding, technologies for the analysis and modification of nucleic acids, next-generation sequencing (NGS), and the bioinformatic approaches required to interpret the results obtained using these methodologies will also be examined.

Detailed program

Cytogenomics
• Banding techniques and advanced methods for the study of cytogenetic abnormalities and the genome
• Fluorescence in situ hybridization (FISH) and its developments
• Array comparative genomic hybridization (array CGH)
• Optical Genome Mapping
Mechanisms Underlying Chromosomal Aberrations
• Chromosomal syndromes and genomic disorders

• Segmental duplications
• Microdeletion and microduplication syndromes
Prenatal Diagnosis: Non-Invasive and Invasive Tests
• Non-invasive prenatal testing (NIPT)
• Chorionic villus sampling
• Amniocentesis
• Mosaicism
UPD and Its Implications in Imprinting Disorders
Cancer Cytogenetics
Molecular Diagnostic Techniques in Medical Genetics
• Simulation of exome analysis and interpretation of genetic variants
• Precision medicine: genetics in support of clinical protocols
• Cancer predisposition, with a focus on acute lymphoblastic leukemia
• Medical genetics: genetic counseling and diagnostic testing
• Non-Mendelian inherited diseases: trinucleotide repeat expansions
Clinical Cases

Prerequisites

See the information provided in the course syllabus

Teaching form

See the information provided in the course syllabus

Textbook and teaching resource

See the information provided in the course syllabus

Semester

See the information provided in the course syllabus

Assessment method

See the information provided in the course syllabus

Office hours

See the information provided in the course syllabus

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | GENDER EQUALITY | REDUCED INEQUALITIES