UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Genetic Pathology

2223-2-H4101D255-H4101D029M

Aims

In-depth study and examples of genetic diseases with Mendelian inheritance and complex inheritance.
Exemplification of the integration of biology and genetics in the formulation and application of personalized and precise clinical protocols
To acquire knowledge about predisposition to cancer and possible clinical application.

Contents

Mendelian genetics: from mutations to examples of pathologies.
Non-Mendelian genetics, complex diseases, multifactorial inheritance.
Precision medicine and cancer genetics, examples of integrated clinical protocols.

Detailed program

Mendelian genetics: mutations and polymorphisms, models of inheritance, family trees. Examples of Mendelian genetic diseases.

Non-Mendelian Genetics: Single-gene defects with non-Mendelian transmission, triplet expansion diseases, epigenetic defects, genomic imprinting, mutations in mitochondrial genes, complex diseases, multifactorial inheritance.

Cancer genetics: precision medicine, risk-based protocols (genetics and response to therapy) in hemato-oncology. Predisposition to cancer, with a focus on leukemia.

Practical examples of Next Generation Sequencing (NGS) analysis

Prerequisites

Bases of genetics and molecular biology

Teaching form

Lectures, with application examples

Textbook and teaching resource

Material provided by the teacher, slides and in-depth articles.

Recommended texts:
Thompson and Thompson, Genetics in medicine
Strachan & Reid, Human Molecular Genetics

Semester

second semster

Assessment method

questions in the examination of the Pathology module

Office hours

on demand by email

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | GENDER EQUALITY