

# UNIVERSITÀ DEGLI STUDI DI MILANO-BICOCCA

## **COURSE SYLLABUS**

## **Genetic Disease**

2122-2-I0301D007-I0301D027M

#### **Aims**

Description of the atypical mechanisms of inheritance - Description of the diseases due to imprinting defects or to dynamic mutations, and mitochondrial and multifactorial diseases - Description of clinical cancer genetics examples

#### **Contents**

By the end of the course, the students will have acquired the general concepts and specific knowledge of ethiopathogenesis of genetic diseases

### **Detailed program**

Classification and incidence of genetic diseases disorders of the autosomes and the sex chromosomes; effects on the phenotype.

Monogenic diseases with Mendelian inheritance and effects on the phenotype; gain and loss of function mutations - Autosomal dominant and recessive inheritance: the concepts of reduced penetrance, variable expressivity, de novo mutation, germline mosaicism - Examples of pathologies - Concepts of clinical heterogeneity, locus heterogeneity, genotype-phenotype correlation Non-mendelian inheritance: 1) unstable repeat expansion diseases (Huntington's and Fragile X syndrome); genetic anticipation; 2) diseases associated with Genomic Imprinting: Angelman and Prader-Willi syndromes; 3) mitochondrial diseases Common diseases: the role of DNA polymorphisms in genetic susceptibility Cancer predisposition syndromes: predisposition to breast and colon cancer. Genetic counseling and

Prerequisites
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Teaching form
Lesson in attendance, subject to any ministerial changes following the COVID pandemic situation
Textbook and teaching resource
ES Tobias; M Connor; M Ferguson- Smith FONDAMENTI DI GENETICA MEDICA Ed. Pearson
Semester
First semester
Assessment method
Exam: quizzes with multiple choice. Test in attendance, subject to any ministerial changes following the COVID pandemic situation
Office hours
On request by e-mail

classification of genetic testing.