



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Patologia Genetica

2122-2-I0302D007-I0302D027M

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#### 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 etiopathogenesis 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

classification of genetic testing.

### **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**

Test in attendance, subject to any ministerial changes following the COVID pandemic situation







## Office hours

On request by e-mail

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