



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Patologia Genetica

1920-2-I0303D007-I0303D032M

---

#### 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 inheritance: the concepts of reduced penetrance, variable expressivity, de novo mutation, germline mosaicism - Autosomal Recessive inheritance: Cystic fibrosis and mutational spectrum - X-linked inheritance: Duchenne and Becker muscular dystrophies, 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**

-

## **Teaching form**

Lectures

## **Textbook and teaching resource**

ES Tobias; M Connor; M Ferguson- Smith FONDAMENTI DI GENETICA MEDICA Ed. Pearson

## **Semester**

First semester

## **Assessment method**

Written exam with multiple choice test, to evaluate global knowledges about course program,

## **Office hours**

On request by e-mail

---