UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Patologia Genetica

1920-2-I0302D007-I0302D027M

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

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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