UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Genetica Molecolare Umana

2223-1-F0601Q104

Aims

The general aim of this course is to introduce the rapid advancements in our understanding the role of human genome in health and disease.
1. Knowledge and understanding
The student will gain knowledge on the rationale of the different approaches used to map disease genes
2. Applying knowledge and understanding
The student will be able to apply the acquired knowledge to the reading of original articles from literature
3. Making judgements
The student will be able to critically discuss and compare different original articles from literature
5. Learning skills
the student will acquire the methodological and scientific skills required to understand and discuss human molecular genetics approaches

Contents

Structure of the human genome
Pedigree and pathogenic mutations
Monogenic and polygenic diseases
Genomic imprinting and dynamic mutations
Linkage analysis
Genome-wide association studies (GWAS)
From the "candidate-gene" to the "disease-gene"
Positive natural selection in the human lineage

Detailed program

Structure of the human genome: the sequencing of the human genome. Genetic variability in humans: characteristics, methods of study and use in genetic mapping. The HapMap project.
Pedigree analysis. Pathogenic mutations in humans. Complex genotype-phenotype correlations: expressivity and penetrance.
Monogenic genetic diseases: examples.
Polygenic diseases: basic concepts and examples. Introduction to quantitative genetics
Genomic imprinting: non-equivalence of maternal and paternal genomes and phenotypic consequences. Examples and interpretations
Linkage analysis: basic concepts, application and examples of use to map disease-genes
Genome-wide association studies (GWAS): basic concepts, applications and examples of use to map variants associated with pathological phenotypes
From the "candidate-gene" to the "disease-gene": design of strategies through examples
Positive natural selection in humans: is it possible to identify regions subject to positive selection? tools, examples and interpretations.

Prerequisites

a solid background in Genetics and Molecular Biology

Teaching form

class lessons

Textbook and teaching resource

The course is based on original articles and online resources that will be indicated when needed

Semester

First semester

Assessment method

oral exam. Students will be asked to choose an original article within a list of selected papers. The discussion of this article will be the starting point of the exam

Office hours

on appointment by e-mail (antonella.ronchi@unimib.it)

Sustainable Development Goals