UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Molecular Genetics Human

2627-1-F0602Q104

Aims

The course aims to introduce approaches to studying the human genome for identifying functional genotype-phenotype relationships in both normal and pathological conditions.
The course objectives for students include:
1.Knowledge and understanding of the basic approaches to mapping disease genes in humans, recognizing their differences and peculiarities.
2.Ability to apply the knowledge acquired in point 1 to read and understand original scientific articles related to the topics covered, and to develop critical thinking in their discussion.
3.Development of methodological and scientific design skills.
4.Development of communication skills: the ability to use the specific language of the discipline to critically discuss the topics covered.
5.Acquisition of the necessary skills to independently pursue further specialized studies in human molecular genetics.

The teaching approach aims to gradually introduce the habit of using the scientific method (ability to define scientific questions, design strategies to answer them, and experimentally validate the answers obtained).

Contents

Structure of the human genome and tools for its study.
Pedigree analysis and pathogenic mutations.
Genomic imprinting and dynamic mutations.
Linkage analysis studies.
Genome-wide association studies (GWAS).
From "candidate gene" to "disease gene".
Genetic history of the human genome.
Positive natural selection in humans.

Detailed program

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

Prerequisites

a solid background in Genetics and Molecular Biology

Teaching form

15 lectures of 2 hours each in a delivery mode: presentation and explanation of content, concepts, and scientific principles.
6 interactive 2-hour lessons:
-Students will be given problems for which theywill asked to design experimental approaches (define the scientific question, design strategies to answer it, and experimentally validate the answers obtained). These approaches will be discussed in class evaluating pros and cons.
-Students will present scientific work of their choice related to the topics of the Course, which will be discussed in class.
In-class lectures.

Textbook and teaching resource

Strachan T. & Read "Human molecular Genetics" (most recent edition)
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.
There are no mid-term exams planned

Office hours

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

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING