UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Medical Genetics

2425-4-H4101D262-H4101D077M

Aims

The module's goals are to help students comprehend the role that genetic tests play in modern diagnosis, the appropriate circumstances under which genetic tests should be ordered, and how to correctly evaluate the findings of genetic and genomic investigations in a clinical setting.

Contents

Main tecniques of cytogenetics, molecular cytogenetics and molecular genetics

Detailed program

• choosing between the different technologies for the diagnosis of genetic diseases; applications, advantages and limitations in comparison: cytogenetics; molecular cytogenetics (FISH and CGH array, SNP array); molecular genetics (PCR, digital and real-time PCR, allele-specific amplification, hybridization with allele-specific oligonucleotides, Sanger and NGS sequencing, MLPA)
• genetic counseling
• genetic tests: classification (diagnostic, presymptomatic, predictive, pharmacogenomics); appropriateness of requests for genetic testing and clinical utility; genetic testing for research; ethical and social aspects of genetic testing; system for genetic variant classification; ability to read and understand information of a genetic report.
• Genetic and Genomic Databases
• genomic diseases and complex diseases
• cancer predisposition syndromes
• invasive and Non invasive prenatal genetic testing
• clinical and labs cases

Prerequisites

Basic science knowledge

Teaching form

Twelve frontal teaching lessons—eleven of which begin with direct instruction and end with interactive teaching with clinical cases, exercises, and FAQs—with the goal of providing a clinical-practical re-elaboration of the concepts presented in the first part of the lesson. One lesson is devoted to interactive teaching on the interpretation of genetic reports.

Textbook and teaching resource

Texbook: Tom Strachan, Andrew Read: Genetica molecolare umana
Slides
Guidelines and scientific articles provided by the teacher

Semester

First semester

Assessment method

Evaluation of knowledges and skills. knowledge gained and reasoning skills on case analyses based on the concepts learned will be assessed.
see syllabus "medicina di laboratorio"

Office hours

Appointemt on request by e-mail
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Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION | GENDER EQUALITY