UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Identificazione di Mutazioni Patogenetiche

2223-1-F0601Q116-F0601Q116M

Aims

The course will be multidisciplinary and organized in modules in order to offer an overview of genetic, molecular, biochemical, physiological and pharmacological aspects of a human disease.
The focus of the course will be the study, from different perspectives, of a genetic disease, from diagnosis to treatment.
In particular, at the end of the course the student will have acquired the following skills:

1. knowledge and understanding: complete overview with different integrated approaches of a specific human genetic disease
2. applied knowledge and understanding: apply what has been learned to the study of numerous human genetic diseases
3. self-judgment: capacity to critically evaluate what has been learned
4. oral competences: oral communication of what has been learned using the correct scientific terminology
5. ability to learn: critical learning and understanding of scientific literature on different aspects of a human disease.

In the academic year 2022-2023 the genetic disease studied will be CYSTIC FIBROSIS.

For the Genetics module (2 CFU), the student will learn how to perform a pedigree analysis; how to identify the disease causing mutation; how to design a prenatal test for a specific disease; how to choose models to study a genetic disease; how to present research papers on the disease under study.

Contents

The course is organized into lectures, group studies and student presentations on specific aspects of the characterization of a genetic disease.

Detailed program

The Genetics module is organized as follows:

- Analysis of the incidence of a specific genetic disease in different populations
- Methods to identify the disease-causing mutation
- Studies of the effects of the disease-causing mutation on gene function
- Analysis and interpretation of the results of scientific research papers on a specific aspect of research on a genetic disease

Prerequisites

Basic knowledge of Genetics.

Teaching form

Lectures, group assignments, students presentations of scientific papers.

Textbook and teaching resource

Powerpoint presentations will be available on e-learning.

Semester

Second.

Assessment method

Written exam.
The exam of the genetics module is part of the general exam of the course that will assess the learning of the material discussed in all the modules.
The written exam is organized in:

1. 30 multiple choice questions on the material discussed in the different modules in 30 minutes
2. 1 open question to answer on the material covered in one module in 15 minutes
   A passing grade on part 1) is required to be able to access part 2) of the exam. The exam will be available on the esamionline platform.

Office hours

Appointment via e-mail with the Genetics module Lecturer.

Sustainable Development Goals

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