UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Identification of Pathogenetic Mutations

2627-1-F0602Q131-F0602Q13103

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 2026-2027 the genetic disease studied will be Cystic Fibrosis.

In the Genetics module (2 CFU), the student will learn how to analyze a genetic tree of a family carrying the disease; how to identify the disease causing mutations; how to identify the cellular and animal models to study the mutation; how to prepare a presentation on a scientific paper and/or on their visit to a lab studying cystic fibrosis.

Contents

The course will be organized into lectures, group studies and student presentations on specific aspects of Cystic Fibrosis.

Visits to research laboratories present in the national territory and specialized in the study of the pathology are planned.

Detailed program

The Genetics module will be subdivided 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 in genetics, molecular biology, biochemistry, physiology and pharmacology.

Teaching form

The course required attendance (10 hours/1 CFU, 2 CFU, 20 hours in total).
It consists in lessons composed by:

a section (2 hours) of delivered didactics (Didattica erogativa, DE) focused on the presentation of contents by the lecturer.
a section (18 hours) of interactive teaching (Didattica Interattiva, DI) including integrative didactic interventions by external experts, group works, reverse teaching with the student's personal involvement.
Visits to centers of excellence in the study of the pathology under consideration are also planned.

Textbook and teaching resource

Please check the main course description.

Semester

Second semester

Assessment method

The verification and evaluation of the student aims to be * innovative * compared to the classic methods.
The exam consists in the elaboration of the knowledges acquired during the course through a poster presentation, similar to what is carried out in scientific meetings.
The student will be evaluated by a commission in terms of: 1) expository skills on what is illustrated in the poster, 2) general knowledge of the pathology treated in the course, 3) first-hand participation in the activities carried out during the course, 4) graphic/display characteristics of the poster.
There are no ongoing tests.

Office hours

Set up an appointment via email (antonella.ronchi@unimib.it, sara.mercurio@unimib.it).

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION