UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Analisi Strutturale della Proteina Mutata

2627-1-F0602Q131-F0602Q13102

Aims

AIMS
The course will have a multidisciplinary and organized in modules providing a comprehensive overview of genetic, molecular, biochemical, physiological and pharmacological aspects of human disease. The focus of the course will be the study, from different perspectives, of a genetic disease, from diagnosis to therapy.
In particular, by 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 learnt to the study of numerous human genetic diseases
3. self-judgement: capacity to critically evaluate what has been learnt
4. communication skills: oral communication of what has been learnt using the correct scientific terminology
5. learning skills: critical learning and undestanding of scientific literature on different aspects of a human genetic diseases.

In the academic year 2025-2026 the human genetic disease studied will be Cystic Fibrosis.

Regarding the biochemistry module (1 CFU), the student will be able to investigate the structure-function relationship of the chlorine channel protein CFTR, assessing the impact of disease-associated mutations on the three-dimensional structure of the protein, in particular the loss of folding and consequent loss of function.

Contents

The course is organized into lectures, group works and interactive teaching on issues concerning structural aspects of the CFTR channel.

Detailed program

The course is structured as follows:

- searching dedicated databases for information on the CFTR protein
- visualisation and analysis of the three-dimensional structure of the wild type protein
- analysis of the impact of disease-associated mutations on protein folding
- analysis of the binding of the mutated protein and the different drugs
- analysis and interpretation of the results of scientific articles related to the topic

Prerequisites

Basic knowledge of biochemistry and computational biology.

Teaching form

The course required attendance (10 hours/1 CFU).
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 (8 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

Powerpoint presentations will be available on e-learning platform.

Semester

Second

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

Appointment via e-mail with the Biochemistry module lecturer (mariaelena.regonesi@unimib.it).

Sustainable Development Goals

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