



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Analisi Strutturale della Proteina Mutata

2223-1-F0601Q116-F0601Q118M

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#### 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 understanding of scientific literature on different aspects of a human genetic diseases.

In the academic year 2022-2023 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 reverse didactics 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 and interpretation of the results of scientific articles related to the topic

## Prerequisites

Basic knowledge of biochemistry and computational biology.

## Teaching form

Lectures, group assignments, students presentations of scientific papers.

## Textbook and teaching resource

Powerpoint presentations will be available on e-learning platform.

## Semester

Second

## Assessment method

Written exam.

The exam of the physiology 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 all the 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 access the part 2) of the exam. The exam will be available on the examonline platform.

## **Office hours**

Appointment via e-mail with the Biochemistry module lecturer.

## **Sustainable Development Goals**

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

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