

SYLLABUS DEL CORSO

Identificazione di Mutazioni Patogenetiche

2425-1-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 2024-2025 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

Please check the main course description.

Textbook and teaching resource

Please check the main course description.

Semester

Second semester

Assessment method

Please check the main course description.

Office hours

Set up an appointment via email.

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION
