

## SYLLABUS DEL CORSO

### Genetica Medica

2324-1-I0302D002-I0302D008M

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#### Aims

The student will learn how :

- to recognize the mode of inheritance of Mendelian characters;
- is the structure of human chromosomes;
- are the mechanisms of mitosis and meiosis
- are the sources of genetic variation;
- are basic concepts of quantitative genetics and population genetics.

#### Contents

The topics of the course concern the basics of formal human genetics, introducing the student to the diagnostic and research approaches of hereditary diseases, including exercises on the probability associated with the inheritance of Mendelian genetic characters

#### Detailed program

- Mitosis and meiosis in relation to conventional cytogenetics.
- Mendelian genetics, extensions, recombination and linkage, genetic and physical maps.
- Mendelian Inheritance in man, pedigree reconstruction.
- Sex determination and X chromosome inactivation.
- Polymorphisms and mutations in the context of genetic variability.
- Basic principles of population and quantitative genetics.

## **Prerequisites**

none

## **Teaching form**

Lectures.

It is required 70% course attendance.

## **Textbook and teaching resource**

Peter J. Russel *Genetica Fondamenti*

Supplementary material will be provided by teacher

## **Semester**

First semester

## **Assessment method**

Being an integrated course, the evaluation will cover all four modules.

Regarding the Medical Genetics module, the evaluation will consist of a written test that will be used to ascertain the level of knowledge and ability to understand the topics covered during the course and to be able to solve problems. The student will have to answer 10 quizzes (Multiple choice test) concerning the topics of Medical Genetics.

Oral examination will be required at professor's discretion (discussion of the written test). The oral test will serve to clarify critical issues emerged from the written test and to verify the communication skills of the student and will focus on the topics covered by the written test.

## **Office hours**

On appointment by e-mail request

# Sustainable Development Goals

GOOD HEALTH AND WELL-BEING

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