



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Modelli Statistici per la Genetica

2324-1-F8203B017

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#### Learning objectives

The course aims to provide fundamental knowledge for a rigorous statistical approach to mapping and identifying loci implicated in human diseases or traits. By the end of the course, students will have learned the essential elements to understand and use basic tools of genetic statistics and methods of genetic epidemiology. Additionally, they will be able to critically read a scientific article and interpret the results derived from statistical analyses of genetic data. Another objective of the course is to equip students with the skills necessary to analyze data from OMIC studies, enabling them to manage and interpret complex genomic datasets.

#### Contents

Mendel vs Complex Diseases: Comparison between simple inheritance and diseases influenced by multiple genes and the environment.

Segregation Analysis: Study of genetic patterns in families.

Parametric and Non-Parametric Linkage: Identification of genomic regions using family or population data.

Genetic Association: Identification of genetic variants through case-control studies.

Sample Size and Power: Number of samples needed to detect significant genetic associations.

GWAS and EWAS: Large-scale studies to find genetic or epigenetic variants associated with traits or diseases.

#### Detailed program

- Mendel and Complex Genetic Diseases
- Hardy-Weinberg Equilibrium
- Factors Complicating Inheritance Identification
- Segregation Analysis

- Linkage Analysis: Theoretical Foundations and Strategies
- Strategies for Genetic Mapping of Mendelian Diseases and Complex Traits
- Parametric and Non-Parametric Linkage Analysis: Methods for identifying genomic regions associated with genetic traits using family information (parametric) or population data (non-parametric: Loss of Heterozygosity, Homozygosity Haplotype Analysis)
- Genetic Association Analysis
- Linkage Disequilibrium: Concepts and Applications
- Case-Control Studies: Methodologies and Analysis
- Family Studies: TDT (Transmission Disequilibrium Test)
- Genome-Wide Analysis in Genetic (GWAS) and Epigenetic (EWAS) Studies: Design and Planning
- Genome-Wide Analysis in Genetic (GWAS) and Epigenetic (EWAS) Studies: Quality Control
- Genome-Wide Analysis in Genetic (GWAS) and Epigenetic (EWAS) Studies: Association Analysis

## Prerequisites

no Prerequisites are needed

## Teaching methods

The course is organized into lectures and practical sessions using dedicated software, aimed at applying the theoretical concepts presented to experimental datasets, as well as interpreting and understanding scientific evidence derived from the correct application of statistical techniques.

## Assessment methods

Written exam (comprising 16 exercises, multiple-choice questions, and open-ended questions on topics covered in class aimed at assessing preparation on the exam syllabus and the ability for independent reflection on critical points of the curriculum).

Optional Oral exam upon request by the student or the instructor (Discussion on topics covered in class and exam texts).

The student must demonstrate not only the ability to reason about correct analysis techniques but also to interpret obtained results and communicate scientifically accurate evidence (problem-solving).

## Textbooks and Reading Materials

Articoli Scientifici ad hoc forniti durante il corso

## Semester

II Semester

## **Teaching language**

Italiano

## **Sustainable Development Goals**

QUALITY EDUCATION

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