



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Genomica Funzionale

2425-1-F0901D040

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#### Aims

The Human Genome Project and subsequent technological advancements, particularly the massive parallel DNA sequencing technologies and techniques for studying the three-dimensional organization of chromatin, have changed the landscape of the relationship between genetics and medicine. The era of medical genetics, focused on chromosomal abnormalities and monogenic diseases, is giving way to the era of clinical genomics and public health. Genome-wide analyses of genetic variability are beginning to comprehensively link the genome to the phenome. Researchers have therefore shifted their focus towards understanding gene functions and the regulatory mechanisms that control gene expression. This includes studying gene-gene and gene-environment interactions to better understand complex traits and diseases.

The aim of the Functional Genomics Course is to acquaint students with the advanced techniques, tools, and experimental strategies used in the post-genomic era for studying the function of gene sequences, their interactions, and the mechanisms regulating their expression. The ultimate goal is to understand and develop the ability to plan genomic studies, which have become a fundamental tool in biomedical research and clinical applications today.

#### Contents

The primary objective of the course is to provide students with an advanced understanding of the functional organization of the human genome. To this end, experimental strategies and the main advanced techniques of genomics and post-genomics will be discussed.

Primary contents: Chromatin and human genome organization; Expression and gene regulation; Epigenetics; Developmental genetics; Mutations, DNA repair and genetic variability; Mapping of variants and disease genes; Evolutionary and population genetics.

The Human Genome Project and subsequent technological developments are to be considered an indispensable

tool for understanding study strategies.

## **Detailed program**

Organization of the human genome; Structure and function of human chromosomes; Model organisms; Comparative genomics and evolution; Genome sequencing; Identification and analysis of functional genome components; Regulation of gene expression in humans; Epigenetics; Non-coding RNAs; Next generation sequencing and its main applications; Single-cell genomic and transcriptomic analysis; DNA methylation analysis techniques; Advanced techniques for chromosomal interaction and three-dimensional chromatin conformation analysis. Genetic variability and its consequences; Strategies for studying gene expression and function; Cellular models, stable cell lines, primary stem cells, and reprogramming; Plasmid and viral expression vectors and applications for studying protein-protein and DNA-protein interactions and potential therapeutic applications; Gene targeting techniques, genome editing, and post-transcriptional modifications for gene deletion or deregulation. Reading and presenting scientific articles.

## **Prerequisites**

Advanced knowledge in Human Genetics and Cellular and Molecular Biology.

## **Teaching form**

- 14 two-hour erogative lectures delivered in-person;
- 2 two-hour interactive lectures delivered in-person;
- 4 two-hour erogative lectures delivered remotely;
- 6 two-hour interactive workshops conducted in-person.

The course will be taught in Italian. The provided teaching materials will be in English.

## **Textbook and teaching resource**

- Lecture slides
- Reviews and articles published in international journals will be indicated during the course.

Recommended Textbooks:

- "Genetica e Genomica nelle scienze mediche"; T. Strachan, J. Goodship, P. Chinnery. Prima Edizione Italiana - Zanichelli
- "Functional Genomics: Methods and Protocols" (Methods in Molecular Biology) edited by Michael Kaufmann, Christine A. Wells, and Athanasios Alexiou.

- "Epigenetics"; Lyle Armstrong. Garland Science

## **Semester**

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## **Assessment method**

Assessment will take place during the scheduled exam sessions through a written exam composed of multiple-choice questions covering the entire course program to evaluate the student's general preparation, and an open question chosen by the student from three proposed questions to assess comprehension ability and in-depth understanding of the topics.

During the course, students will also be prompted to prepare an oral presentation on a topic of their choice relevant to the program, to evaluate their presentation and synthesis skills.

The final grade will be determined by the average obtained from the scores obtained in the multiple choice test and the open question.

Upon request by the professor or the student, a brief oral exam may be conducted, consisting of an interview on the topics covered in class and/or a discussion of the written exam.

## **Office hours**

on appointment, by e-mail arrangement ([emanuele.azzoni@unimib.it](mailto:emanuele.azzoni@unimib.it))

## **Sustainable Development Goals**

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

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