UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Functional Genomics

2526-1-F0902D001

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.

Specific Objectives

1. Knowledge and understanding
   Students will acquire an in-depth understanding of functional genomics technologies and experimental approaches, with a focus on high-throughput DNA sequencing, chromatin 3D structure analysis, and gene expression regulation. They will also gain knowledge on genetic variability and gene-gene and gene-environment interactions in the determination of complex traits.

2. Applying knowledge and understanding
   Students will be able to design, conduct, and interpret experimental studies in the field of functional genomics, critically selecting the most appropriate methodologies. They will be capable of applying this knowledge to advanced biomedical research and clinical settings in translational genomics.

3. Making judgements
   Students will develop critical thinking skills in the analysis of genomic data, independently assessing the reliability and significance of scientific results, formulating coherent hypotheses, and evaluating the ethical, clinical, and scientific implications of their analyses.

4. Communication skills

Students will gain the ability to clearly, effectively, and professionally communicate the content and results of complex genomic analyses, both in academic contexts and within multidisciplinary teams, using appropriate scientific terminology and communication tools.

5. Learning skills
   Students will develop a high degree of autonomy in learning and continuous updating, acquiring the ability to critically access scientific literature and independently explore technological and methodological innovations in functional genomics.

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 diseases 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 & Genomica nelle scienze mediche"; Tom Strachan, Anneke Lucassen. Seconda Edizione Italiana - Zanichelli
- "Genetica Molecolare Umana"; Tom Strachan, Andrew Read. Seconda edizione italiana condotta sulle 5 edizione inglese - Zanichelli
- "Functional Genomics: Methods and Protocols" (Methods in Molecular Biology); Michael Kaufmann, Christine A. Wells, Athanasios Alexiou.

Semester

I Semester

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 abilty and in-depth understanding of the topics.

During the course, students will also be prompted to prepare an oral presentation (optional) 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. The optional presentation can contribute to the final grade by adding a maximum of 3 additional points.
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)

Sustainable Development Goals

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