UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Metodologie Bioinformatiche

2526-1-F0803Q054

Aims

ELearning Objectives
Knowledge:
• To understand the main bioinformatic methodologies for the analysis of biological data, particularly omics data.
• To comprehend the theoretical foundations and algorithms underlying bioinformatic analysis tools.
Skills:
• To use bioinformatic tools for processing and interpreting biological data.
• To select and apply appropriate methodologies based on the specific biological question.

Expected Learning Outcomes
At the end of the course, the student will be able to:
• Knowledge and understanding: describe the fundamental principles of major bioinformatic methodologies, with reference to applications in genomics, transcriptomics, and omics data analysis.
• Applying knowledge and understanding: use software environments and tools for the bioinformatic analysis of real datasets, critically interpreting the results obtained.
• Making judgements: assess the appropriateness of different methodological approaches for specific biological or clinical questions, and propose alternative solutions where needed. This skill is developed through interactive activities in class, including guided discussion of case studies, interpretation exercises based on real data, and practical examples worked out collectively.
• Communication skills: clearly and coherently communicate methodological choices and analysis results, both in written form and through oral presentations.
• Learning skills: independently deepen their knowledge of bioinformatic tools and resources not covered in class, keeping up to date through scientific literature and technical documentation.

Contents

• Introduction to bioinformatics
• Pills of Statistics and Artificial Intelligence
• Data generation: from sequencing platforms to genome assembly and annotation, to multi-omics
• Biological data organization and management
• Sequence Analysis
• Transcriptomic data analysis
• Multi-omics data integration

Detailed program

1. Introduction to bioinformatics
2. Pills of Statistics and Artificial intelligence
   a Elements of statistics for omics data analysis
   b. Supervised Machine Learning
   c. Unsupervised Machine Learning
3. Data generation
   a. DNA sequencing platforms
   b. From chromatogram to reads: “base calling”
   c. Coverage, reads quality, data formats
   d. From reads to the sequence: genome assembly
   e. Genome annotation
4. Sequence analysis
   a. Local and global alignment
   b. Exact and heuristic algorithms
5. Biological data organization and management
   a. Databases and DBMS: relational and flat file databases
   b. Biological databases
   i. Genomic databases (GenBank - ENA – DDBJ)
   ii. Proteomic dabases (UniprotKB, Swiss-Prot, TrEMBL – PDB)
   iii. Genome browsers: ENSEMBL, UCSC
6. Analysis of the transcriptome
   a. From RNA sequencing to gene expression levels
   b. Single-cell RNA sequencing data pre-processing and analysis
7. Multi-omics data integration
   a. Enrichment analysis and integrated visualization of transcriptomic, proteomic, and metabolomic data
   b. Integration of omics data into metabolic network models

Prerequisites

Concepts of cellular and molecular biology, and biochemistry provided in basic courses of bachelors in Biological or Biotechnological sciences will be given for granted.
Previous knowledge that is useful, which will be only briefly revised during the course:
• Elements of computer’s architecture and algorithms
• Elements of probability and statistics

Teaching form

42 hours of lectures, divided as follows:
– 36 hours in delivered mode (face-to-face), supported by in-class presentations;
– 6 hours in interactive mode, dedicated to the guided development and discussion of students' final project proposals.

10 hours of practical sessions held in person, focused on the use of biological databases and bioinformatic analysis platforms.

Textbook and teaching resource

Slides and video recordings of lectures and practical sessions will be available on the course e-learning page.
Suggested textbook: Citterich, Ferré, Pavesi, Romualdi, Pesole. Fondamenti di Informatica. BIOLOGIA ZANICHELLI
Specialized research articles, surveys and book chapters will be recommended during the course.

Semester

First semester

Assessment method

Individual or group research project on a topic chosen by the student which involves the critical use of some tools seen in class
+
Oral exam in which the student presents the research project and demonstrates mastery of the concepts used in the project. During the oral exam, sequence alignment exercises (to be carried out on paper) will also be proposed.

There are no intermediate tests for attending students.

Office hours

Students are invited to contact the teacher by email to agree upon a date (possibly on Webex)

Sustainable Development Goals