



UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Long read sequencing of entire genomes and transcriptomes

2425-117R-GENOMES

Aims

The course is focused on the characteristics and applications of third-generation sequencing technologies - leveraging long reads. Particular emphasis will be devoted to Nanopore-based technologies.

Knowledge and understanding.

Students will familiarize with the main technologies for the sequencing of nucleic acids, deepening those that enable the sequencing of long DNA and RNA molecules. Eventually, they will acquire key concepts regarding the application of these technologies for the characterization of complex genomic regions and their usefulness in the fields of biotechnology and biomedicine.

Applying knowledge and understanding.

Students will be able to apply the acquired knowledge in laboratory experiences and to use the comprehension skill in subsequent studies and/or research activities.

Making judgments.

Students will be able to process what they have learned and be able to recognize situations and problems in which the acquired knowledge can be used.

Communication skills.

At the end of the course, students will be able to express themselves appropriately in the description of issues related to technologies for the sequencing of nucleic acids.

Learning skills

At the end of the course, students will be able to survey the scientific literature related to the topics covered and will be able to analyse, apply and integrate the acquired knowledge with what learned in related courses and research activities.

Contents

1. Intro on sequencing
2. Nanopore sequencing
3. Analysis of genomes
4. Analysis of transcriptomes
5. DNA and RNA modifications
6. Targeted sequencing
7. Protein sequencing
8. Applications

Detailed program

1. Intro on sequencing: (i) Background on HTS, (ii) Limits of short-reads sequencing, (iii) Short- vs long-reads, (iv) Key long-reads technologies
2. Nanopore sequencing: (i) Key concepts, (ii) Platforms, (iii) Key data analysis concepts, (iv) Mapping/Alignment algorithms
3. Analysis of genomes: (i) Genome assembly, (ii) High-quality assembly of complex plant genomes, (iii) Human population studies, (iv) Microbial communities: metagenomics
4. Analysis of transcriptomes: (i) Isoform level / alternative splicing analysis, (ii) Identifying full length transcripts, (iii) Transcriptional diversity and annotation, (iv) Single-cells, (v) Spatial transcriptomics
5. DNA and RNA modifications
6. Targeted sequencing
7. Protein sequencing
8. Applications

Prerequisites

Background: basic notions of Molecular Biology.

Prerequisites: none

Teaching form

4 x 2 hours-lectures of delivered didactics (Didattica erogativa, DE) focused on the presentation-illustration of contents by the lecturer.

Didactic activities are conveyed by means of face-to-face lectures.

Teaching language: Italian.

Textbook and teaching resource

Slides, and recording of the lessons will be available at the e-learning platform of the course. Specific literature is reported within the slides.

Semester

Second semester

Assessment method

Oral examination. Students will be asked to discuss the topics covered during the course.

Office hours

By appointment through e-mail request to the lecturer.

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING
