

General information	
Academic subject	Bioinformatica ed analisi del genoma
Degree course	LAUREA MAGISTRALE IN BIOTECNOLOGIE PER LA QUALITÀ E LA SICUREZZA DELL'ALIMENTAZIONE
Academic Year	I
European Credit Transfer and Accumulation System (ECTS)	Total CFU: 6, including frontal lesson CFU: 5 ese/lab/tutor CFU:1
Language	Italian
Academic calendar (starting and ending date)	October 3, 2022 - January 27, 2023
Attendance	Not mandatory

Professor/ Lecturer	
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Virtual headquarters	
Tutoring (time and day)	Every day, from Monday to Friday, upon agreement by e-mail

Syllabus	
Learning Objectives	<p>The main purposes of the teaching program, reported below, are:</p> <ul style="list-style-type: none"> - Acquisition of skills on common databases, algorithms and tools for bioinformatic analysis. - Acquisition of knowledge about the fundamental structural characteristics and functions of prokaryotes and eukaryotes genomes. - Acquisition of knowledge about the main bioinformatic resources for the exploration and analysis of genomes and their products. - Learning of the main bioinformatics tools for comparative genomics and metagenomics studies.
Course prerequisites	Molecular biology, biochemistry, and genetics basic knowledge.
Contents	<p>1) Basic concepts of bioinformatic method and introduction to -omics biology</p> <ul style="list-style-type: none"> - Elements of bioinformatics: databases, algorithms and software for the management and analysis of biomolecular data. - Analysis of genome and its products; concepts and analysis of structural, functional and comparative genomics. <p>2) The prokaryotic genome</p> <ul style="list-style-type: none"> - Structure, dimension, plasticity, base composition and functional content of prokaryotic genomes. - Organization of prokaryotic genes and operons. - Mobile elements and pathogenicity islands. - Prokaryotes genetic code. - Small non-coding RNA.



3) Taxonomic identification based on genomic markers

- Molecular and bioinformatic methods for biodiversity study.
- DNA barcoding: genomic markers of species, barcoding gap, reference databases for the molecular identification of species.
- Common biomolecular and bioinformatics protocols in DNA barcoding analysis.

4) Molecular and bioinformatic protocols of metagenomic analysis

- Microbiome analysis: metagenomic DNA extraction, sequencing, assembly and binning. Functional annotation, comparative analysis, multi-sample statistics and data integration/sharing.
- Molecular and bioinformatics tools for DNA metabarcoding analysis: Next Generation Sequencing data production; bioinformatic pipelines and reference databases for the microbiome taxonomic profiling; example: the pipeline BioMaS.
- Molecular and bioinformatic tools for shotgun metagenomic analysis: Next Generation Sequencing (NGS) data production; bioinformatics pipelines and reference databases for the microbiome taxonomic and functional characterization; common approaches to human metagenome analysis.
- Examples of real metagenomic datasets: use of EBI – Mgnify bioinformatic resource; example of microbiome taxonomic, statistical and comparative analysis in relation to diet through Qiime tool.

5) The eukaryotic genome

- Structural and functional features of eukaryotic genome: dimension, chromosomes organization, DNA content, sequence complexity, synteny, euchromatin and heterochromatin portions, gene density.
- Compositional features and gene expression regulation: base composition, isochores model, epigenetic regulation of gene expression, CpG islands; a modern definition of gene.
- Informational composition of the eukaryotic genome (the human genome case study): coding, non-coding, unique and repeated sequence fractions overview. Gene families. Monocistronic and polycistronic genes.
- Structure and dimensions of eukaryotic genes and their functional domains; mRNA processing; trans-splicing molecular mechanism; alternative splicing and transcripts iso-orthology; non-coding RNAs analysis: rRNA, tRNA, snRNA, snoRNA, microRNA, lncRNA, antisense RNA and circular RNAs.
- Gene families and pseudogenes: structure, function, evolution and analysis of gene families and pseudogenes and their role in the eukaryotic genome plasticity.
- In tandem and interspersed repeated non-coding elements: segmental duplications, microsatellites, minisatellites, retrotransposons, DNA transposons.

6) The mitochondrial genome

- mtDNA size, shape and informational content.
- Base composition, genetic code, transcripts editing.
- Overview of yeast, plants and metazoans mitochondrial genomes.
- Bioinformatic and evolutionary analysis of mtDNA and its use in population and clinical genetics studies.

7) Genomic sequencing and bioinformatic analysis of produced Big Data

- Whole genome shotgun sequencing, assembly and bioinformatic analysis.

	<ul style="list-style-type: none"> - Sequencing quality score. - Common Next Generation Sequencing platforms. <p>8) Bioinformatics hands-on</p> <ul style="list-style-type: none"> - Primary and specialized databases: structure, content and retrieval systems. - Database similarity searching. - Genomic Browsers: examples, query systems and similarity searching. - Functional and structural characterization of genomic sequences. - Taxonomic analysis of microbiomes based on DNA metabarcoding data.
Books and bibliography	<ul style="list-style-type: none"> - Fondamenti di bioinformatica. Manuela Helmer Citterich, Fabrizio Ferrè, Giulio Pavesi, Graziano Pesole, Chiara Romualdi. Zanichelli - Biologia Molecolare. Francesco Amaldi Piero Benedetti Graziano Pesole Paolo Plevani. Terza edizione. Casa Editrice Ambrosiana.
Additional materials	It is advisable to integrate the consultation of the suggested books with the study of lesson notes and slides made available on the Teams platform in the “BIOINFORMATICA ED ANALISI DEL GENOMA” team.

Work schedule			
Total	Lectures	Hands on (Laboratory, working groups, seminars, field trips)	Out-of-class study hours/ Self-study hours
Hours			
52	40	12	98
ECTS			
6	5	1	
Teaching strategy	Lectures supported by PowerPoint slides and practical exercises on the computer.		
Expected learning outcomes			
Knowledge and understanding on:	<ul style="list-style-type: none"> ○ Acquisition of theoretical and practical knowledge in the field of bioinformatics. ○ Acquisition of basic knowledge concerning the genomic structure, functions and analysis. 		
Applying knowledge and understanding on:	<ul style="list-style-type: none"> ○ Knowledge of the main bioinformatics tools for the analysis of genomic sequences. ○ Ability to apply basic bioinformatic methodologies for genomic analysis. 		
Soft skills	<ul style="list-style-type: none"> • <i>Making informed judgments and choices</i> <ul style="list-style-type: none"> ○ Ability to critically approach molecular and bioinformatic analytical protocols, by evaluating their advantages and disadvantages related to specific case studies in order to select the most suitable methodology and set its parameters correctly. • <i>Communicating knowledge and understanding</i> <ul style="list-style-type: none"> ○ Acquisition of suitable terminology for the description of -omics and bioinformatics methodologies. • <i>Capacities to continue learning</i> <ul style="list-style-type: none"> ○ Acquisition of a learning method targeted at biomolecular and bioinformatics analysis. 		

	<ul style="list-style-type: none"> ○ Acquisition of the required knowledge for autonomous and targeted selection of data and tools for genomic analysis.
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Assessment and feedback	
Methods of assessment	Mid-time oral exam, final oral exam.
Evaluation criteria	<ul style="list-style-type: none"> • <i>Knowledge and understanding</i> <ul style="list-style-type: none"> ○ Understanding of the fundamental concepts underlying the structural, functional and comparative analysis of genomes. • <i>Applying knowledge and understanding</i> <ul style="list-style-type: none"> ○ Skills in common biomolecular databases and bioinformatic tools use and ability to interpret their results. • <i>Autonomy of judgment</i> <ul style="list-style-type: none"> ○ Critical ability in illustrating the acquired knowledge on common tools for the genomes study. Ability to build logical paths in setting up a bioinformatics analysis workflow. • <i>Communicating knowledge and understanding</i> <ul style="list-style-type: none"> ○ Clear and logical description of the basic concepts of -omic biology and bioinformatics. • <i>Communication skills</i> <ul style="list-style-type: none"> ○ Use of correct terminology, suitable to the topic. • <i>Capacities to continue learning</i> <ul style="list-style-type: none"> ○ Understanding of the fundamental concepts of genomics and bioinformatics aimed at correct choice and use of analysis tools.
Criteria for assessment and attribution of the final mark	The learning assessment and the final mark attribution will be carried out based on the criteria detailed in the didactic regulation of the degree course (art.6).
Additional information	