Universitat Autònoma de Barcelona

2021/2022

Genomics

Code: 42399
ECTS Credits: 12

Degree	Type	Year	Semester
4313473 Bioinformatics	OT	0	1

The proposed teaching and assessment methodology that appear in the guide may be subject to changes as a result of the restrictions to face-to-face class attendance imposed by the health authorities.

Contact

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Use of Languages

Principal working language: english (eng)

Teachers

Marta Coronado Zamora
Jaime Martínez Urtaza
Juan Ramón González Ruíz
Sònia Casillas Viladerrams
Raquel Egea Sánchez

External teachers

Miguel Pérez-Enciso
Sergi Hervás
Simon Heath
Yolanda Guillén
Óscar Lao

Prerequisites

To carry out this module is necessary to have passed previously both compulsory modules: Programming in Bioinformatics and Core Bioinformatics. Basic notions in genetics are also needed.

It is recommended you have a Level B2 of English or equivalent.

Objectives and Contextualisation

The technological capacity to generate massive genomic and multiomics data grows at a relentless pace without a parallel growth of the bioinformatics expertise to deal with the integration of molecular data.

The purpose of this module is to provide the knowledge and technical skills which are required to successfully meet the current challenges of genomic and multiomics analyses.

Competences

- Analyse and interpret data deriving from omic technology using biocomputing methods .
- Communicate research results clearly and effectively in English.
- Design and apply scientific methodology in resolving problems.
- Identify the biocomputing needs of research centres and companies in the biotechnology and biomedicine sectors.
- Possess and understand knowledge that provides a basis or opportunity for originality in the development and/or application of ideas, often in a research context.
- Propose biocomputing solutions for problems deriving from omic research.
- Propose innovative and creative solutions in the field of study
- Understand the molecular bases and most common standard experimental techniques in omic research (genomics, transcriptomics, proteomics, metabolomics, interactomics, etc.)
- Use and manage bibliographical information and computer resources in the area of study
- Use operating systems, programs and tools in common use in biocomputing and be able to manage high performance computing platforms, programming languages and biocomputing analysis.

Learning Outcomes

1. Communicate research results clearly and effectively in English.
2. Describe and apply the tools of assembly, annotation, storage, display and analysis of the variation in genomes.
3. Describe the operation, characteristics and limitations of first, second and third generation sequencing techniques.
4. Design and apply scientific methodology in resolving problems.
5. Design and interpret studies of association between genetic polymorphisms and phenotype for identifying genetic variants affecting phenotype characters, including those associated pathologies and that confer susceptibility to human diseases or other species of interest.
6. Enumerate and describe the content of databases of relevant information for the different areas of genomics and carry out advanced searches.
7. Establish the corresponding relationships between nucleotide sequencing, structure and gene function using sources of biological data and the bases of biocomputing analysis.
8. Identify and characterize sources and display formats genomes, along with your notes and information about genetic variation, disease association and gene expression.
9. Integrating genomic data in silico reconstruction of cells and organisms (systems biology, synthetic biology).
10. Possess and understand knowledge that provides a basis or opportunity for originality in the development and/or application of ideas, often in a research context.
11. Propose innovative and creative solutions in the field of study
12. Recognise the strategic importance of genetic advances in the area of human health, especially the genomic applications in personalised medicine and pharmacogenomics.
13. Use and manage bibliographical information and computer resources in the area of study
14. Use the latest algorithms of alignment of sequences and generation of evolutive trees as well as sequencing methods and gene prediction.

Content

Lesson 1. Introduction: Genome and Omics data

Lesson 2. Next Generation Sequencing (NGS)

Lesson 3. Primary NGS data analysis

Lesson 4. Making sense of genome data

4.1 Genome assembly

4.2 Genome annotation

4.3 Functional analysis

Lesson 5. Genome Visualization

Lesson 6. Genome variation

6.1 Theory and data

6.2 Nucleotide variation

6.3 Structural variation

Lesson 7. Association and GWA studies

Lesson 8. Transcriptomics

8.1 Microarrays

8.2 RNAseq

Lesson 9. Systems Genetics: Omics data integration

Lesson 10. Metagenomics

Lesson 11. Single cell

Lesson 12. Applied genomics: solving problems and real cases

Student Seminars' Session

Closing Conference

*Unless the requirements enforced by the health authorities demand a prioritization or reduction of these contents.

Methodology

The methodology combines master classes, solving practical problems and real cases, work in the computing lab, performing individual and team works, readings and discussing papers related to the thematic blocks. As ICT resource we will use the virtual teaching platform of the master.

*The proposed teaching methodology may experience some modifications depending on the restrictions to face-to-face activities enforced by health authorities.

Annotation: Within the schedule set by the centre or degree programme, 15 minutes of one class will be reserved for students to evaluate their lecturers and their courses or modules through questionnaires.

Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Seminars	4	0.16	
Solving problems in class and work in the computing lab	28	1.12	
Theoretical-Practical classes	37	1.48	

Type: Supervised		
Performing individual and team works	120	4.8
Type: Autonomous		
Regular study	107	4.28

Assessment

- Work done and presented by the student (student's portfolio) (55%).
- Individual theoretical and practical tests (35%)
- Soft skills (assistence, arrival on time and proactive participation in class) (10%)

The student will be graded as "No avaluable" if the weight of the evaluation is less than 67% of the final score. The teaching staff will inform about the procedures and deadlines for the retake process. Note that soft skills cannot be retaken.

To be eligible for the retake process, the student should have been previously evaluated in a set of activities equaling at least two thirds of the final score of the module

**Student's assessment may experience some modifications depending on the restrictions to face-to-face activities enforced by health authorities.*

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Individual theoretical and practical test	35%	4	0.16	1, 3, 2, 4, 5, 6, 7, 8, 9, 11, 12, 10, 14, 13
Soft skills (assistence, arrival on time and active participation in class)	10%	0	0	1, 4, 11, 10, 13
Student's portfolio	55%	0	0	1, 3, 2, 4, 5, 6, 7, 8, 9, 11, 12, 10, 14, 13

Bibliography

Basic references

- Compeau, P and P. Pevzner. 2015. Bioinformatics Algorithms Volume 1 and 2. 2n edition. Active Learning Publishers LLC
- Gibson, G. i S. V. Muse, 2009. A Primer of Genome Science. Sinauer, Massachusetts. 3rd edition.
- Barnes, M. 2007. Bioinformatics for geneticists (2nd Ed.) Wiley.
- Brown, T. A. 2018. Genomes. 4th edition. Taylor & Francis Inc.
- Lesk, M. K. 2017. Introduction to Genomics. 3rd edition. Oxford Univ. Press.
- Makinen, V.; A. Belazzougui, F. Cunial, A.I. Tomescu. 2105. Genome-Scale Algorithm Design: Biological Sequence Analysis in the Era of High-Throughput Sequencing. Cambridge Univ Press.
- Pevnser, J. 2009. Bioinformatics and Functional Genomics (2nd edition). Wiley-Blackwell.
- Pevzner, P. and R. Shamir. 2011. Bioinformatics for Biologists. Cambridge University Press
- Samuelsson, T. 2012. Genomics and Bioinformatics: An Introduction to Programming Tools for Life Scientists. Cambridge University Press.

Recomended Websites

- Course: Gurrent topics in Genome Analysis 2016. NHGRI (http://www.genome.gov/12514288)
- National Human Genome Research Institute (USA) (http://www.genome.gov/)
- Genomic careers (http://www.genome.gov/genomicCareers/video_find.cfm)
- 1000 genomes project (http://www.internationalgenome.org/)
- PopHuman database (http://pophuman.uab.es)
- Genome online databases (GOLD) (https://gold.jgi.doe.gov/)
- Genome data viewer NCBI (https://www.ncbi.nlm.nih.gov/genome/gdv/)
- Ensembl genome browser (http://www.ensembl.org)
- UCSC genome browser (http://genome.ucsc.edu/)
- Genome size databases (http://www.genomesize.com/)

Software

Software to be used through the module

- R https://cran.r-project.org/
- Rstudio https://www.rstudio.com/products/rstudio/
- Fastqc https://www.bioinformatics.babraham.ac.uk/projects/fastqc/
- bwa http://bio-bwa.sourceforge.net/
- vcftools https://github.com/vcftools/vcftools/zipball/master
- bedtools https://bedtools.readthedocs.io/en/latest/
- GATK https://software.broadinstitute.org/gatk/
- IGV https://software.broadinstitute.org/software/igv/
- JBrowse https://jbrowse.org/jb2/