

## Current Topics in Bioinformatics

Code: 105065  
ECTS Credits: 3

**2024/2025**

Degree	Type	Year
2500890 Genetics	OT	4

## Contact

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## Teachers

Marta Coronado Zamora

## Teaching groups languages

You can view this information at the [end](#) of this document.

## Prerequisites

- It is recommended to have passed *Bioinformatics* (3rd year), *Genomics*, *Proteomics and Interactomics* (3rd year) and the module of *Databases and programming fundamentals* within *Instrumental Techniques* subject (2nd year) of the Degree in Genetics.
- Basic knowledge of a programming language and familiarity with the Linux environment are essential to follow the practical sessions and complete the continuous assessment activities.
- A level B1.2 of English or equivalent is recommended.

## Objectives and Contextualisation

The purpose of this course is to cover basic topics in bioinformatics through practical workshops, seminars, and lectures given by experts in the field. It is not a cumulative course but rather a transversal one, aiming to provide students with a broad range of concepts and approaches encompassed by bioinformatics.

The main objective is to equip students with the necessary knowledge and skills to apply bioinformatics in various areas of genomic research and other omics disciplines. The material taught and the activities conducted during this course offer a comprehensive perspective on the potential of bioinformatics in both basic and applied research.

## Competences

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
- Be able to analyse and synthesise.
- Be able to communicate effectively, orally and in writing.
- Describe and identify the structural and functional characteristics of nucleic acids and proteins including their different organisational levels.
- Describe the organisation, evolution, inter-individual variation and expression of the human genome.
- Develop self-directed learning.
- Know and apply the omic' tools of genomics, transcriptomics and proteomics.
- Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
- Measure and interpret the genetic variation in and between populations from a clinical, conservational and evolutionary perspective, and from that of the genetic improvement of animals and plants.
- Perceive the strategic, industrial and economic importance of genetics and genomics to life sciences, health and society.
- Reason critically.
- Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
- Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
- Use and interpret data sources on the genomes and macromolecules of any species and understand the basics of bioinformatics analysis to establish the corresponding relations between structure, function and evolution.
- Use and manage bibliographic information or computer or Internet resources in the field of study, in one's own languages and in English.

## Learning Outcomes

1. Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
2. Be able to analyse and synthesise.
3. Be able to communicate effectively, orally and in writing.
4. Defend the relevance of progress in the generation and interpretation of data on a genomic scale for our understanding and technological manipulation of organisms.
5. Develop self-directed learning.
6. Explain and apply the methods for the analysis and annotation of genomes.
7. Explain how knowledge of human genetic variation is applied to personalised medicine, pharmacogenomics and nutrigenomics.
8. List and explain the content of bioinformatics databases and perform searches for information.
9. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
10. Reason critically.
11. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
12. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
13. Use and interpret the results of bioinformatics applications in the molecular analysis of sequences.
14. Use and manage bibliographic information or computer or Internet resources in the field of study, in one's own languages and in English.
15. Use bioinformatics techniques and tools to describe and analyse the human genome.
16. Use the techniques, tools and methodologies used to describe, analyse and interpret the enormous amounts of data produced by high performance technologies.

## Content

The subject will be composed of theoretical-practical sessions, lectures and workshops given by recognized specialists in the different subjects and fields.

#### Theoretical-practical sessions (~12h)

They will take place in the computer room. Students will work both individually and as a group (3-4 students) promoting active learning that will allow them to develop the capacity for analysis and synthesis, critical reasoning and the capacity to solve problems.

We are about to start a journey into real Bioinformatics. This series of four hands-on training activities will show you the basic workflows in bioinformatics: from the data management and processing with Linux, visualization, and posterior functional analyses. The practicals are divided into two big parts: Part I: Basics on Bioinformatics workflows and Part II: Solving real cases in genomics.

These practicals pretend also to gain other skills, very valuable in research but rarely experienced during the Degree, such as collaborating, learning how to transform the data into effective graphs to communicate and doing reproducible research.

Title		Description and learning outcomes
Introduction		Subject presentation: organization, methodology, preparations, group creation, subject grading
P1. Basics on Bioinformatics workflows	Data management and processing	Learning Linux for Bioinformatics - Learning to manage raw data with basic commands of bash, a powerful Linux language.
	Data exploration and visualization	Data exploration and visualization - Learning how to represent biological data into knowledge graphs. We will use ggplot2, an R package.
P2. Solving real cases in genomics	Machine learning	Application of machine learning - Perform a machine learning analysis in Python.
	Transcriptomic analyses	Finding differential expressed genes in diseases - Perform a differential expression analyses in R.
Mentoring*		*Two extra sessions of two hours will be implemented to be set according to the needs of the students and the difficulty of the cases.

#### Invited Conferences and Workshops (17 hours)

Attendance to the invited conferences (1-2 hours per conference) by invited experts in the field of bioinformatics is mandatory and requires in-person participation. The conferences will be conducted in English and will cover various topics related to the practical application of bioinformatics and genetics in different domains such as hospitals, private companies, and academic institutions. They will also address current and specific topics like cancer research. The exact dates of the conferences will be updated on the calendar and communicated through the communication tools available on the Moodle platform. The conferences will be virtual and may be recorded.

## Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Lectures	10	0.4	4, 6, 7, 8
Theoretical-practical sessions	12	0.48	2, 3, 4, 5, 6, 7, 8, 10, 13, 14, 15, 16
Workshops	7	0.28	3, 5, 6, 10
Type: Supervised			
Portfolio	20	0.8	2, 3, 4, 5, 6, 7, 8, 10, 13, 14, 15, 16
Type: Autonomous			
Study/Problem solving	25	1	5, 8, 10, 13, 15, 16

### In-person learning activities and autonomous learning

A cooperative learning experience will be implemented. Each group must manage and solve practical cases autonomously.

The members of each group will know in depth the information they manage. Each group will make a lecture and/or writing of a portfolio through which the other groups understand the characteristics and foundations of each analysis. The four practical sessions will be linked, since the results or the methodology of one practical will be used for the next practical.

The active participation, the management of the work, as well as the discussion of the acquired knowledge will form a vital part in the role played by each student.

### Conferences and workshops

A total of 5-6 conferences will be delivered by experts in their respective research or professional fields, offering a realistic perspective on bioinformatics as a key component in addressing both basic and applied biological research questions. Emphasis will be placed on the importance of data analysis in the current era of big data. Other conferences will cover practical aspects such as navigating through a Ph.D., science communication using real-life examples, or understanding the scientific world. Additionally, various career paths in bioinformatics within academic and private sectors will be explored.

This course does not have a single evaluation system for the 2024/2025 academic year.

\*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.

## Assessment

## Continuous Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Portfolio	70%	0	0	1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16
Seminar lecture	20%	1	0.04	1, 3, 4, 6, 7, 9, 10, 11, 12, 13
Soft skills	10%	0	0	2, 3, 4, 5, 6, 7, 10, 14

The grading will be carried out through the delivery of four portfolios and a seminar about a bioinformatics topic chosen by the students.

Portfolio (70%). In each portfolio the basic fundamentals of the analyzed data will be exposed, the tools used, the development of the methodology, as well as a discussion on the final result of the delivery. Each portfolio will have the same weight in the final evaluation.

Seminar presentation (20%). Each group will make a 15-minute oral presentation.

Assistance and participation (10%).

The subject is passed when the average score of the assessment activities is equal to or greater than 5. Due to the continuous and transversal nature of this evaluation, the subject cannot be assessed if student participation is below 80% of the proposed sessions.

This course does not have a single evaluation system for the 2024/2025 academic year.

## Bibliography

### Books

- 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
- Lesk, A. 2014. Introduction to bioinformatics. Oxford University Press
- Claverie, J-M. 2007. Bioinformatics for dummies. Wiley, cop
- Hadley, W. 2009. ggplot2: elegant graphics for data analysis. Springer

### Articles

- [Himmelblau, E. \(2021\) A cartoon guide to bioinformatics by a novice coder. Nature](#)
- [Markowetz, F. \(2017\) All biology is computational biology. PLoS Biology 15\(3\):e2002050](#)
- [Webb, S. \(2018\) Deep learning for biology. Nature 554:555-557](#)
- [Mardis, E.R. \(2010\) The \\$1,000 genome, the \\$100,000 analysis? Genome Medicine 2:84](#)
- [Tyler-Smith C, Yang H, Landweber LF, Dunham I, Knoppers BM, Donnelly P, et al. \(2015\) Where Next for Genetics and Genomics? PLoS Biology 13\(7\):e1002216](#)
- [Tippmann, S. \(2015\) Programming tools: Adventures with R. Nature 517, 109-110](#)
- <https://www.biostarhandbook.com/>

### Links

- [Web MSc in Bioinformatics UAB](#)
- [Bioinformatics salaries](#)
- [Bioinformatics: What? Why? Who?](#)

## Software

- Operative system: Linux
- Programming languages: bash, R
- Software: RStudio y Jupyter Notebook
- R packages: ggplot2, shiny, rmarkdown, knitr, BiocManager, DESeq2, clusterProfiler, dplyr, biomaRt, ggrepel, pheatmap, org.Hs.eg.db, pathview
- Python packages: numpy, pandas, matplotlib, requests, sklearn, tensorflow, itertools, seaborn

All the software is installed on the computers at the faculty.

## Language list

Name	Group	Language	Semester	Turn
(PLAB) Practical laboratories	641	Spanish	first semester	morning-mixed
(SEM) Seminars	641	Spanish	first semester	afternoon
(TE) Theory	64	Spanish	first semester	morning-mixed