

**Bioinformatics**

Code: 102890  
ECTS Credits: 3

Degree	Type	Year	Semester
2502442 Medicine	OT	3	0
2502442 Medicine	OT	4	0
2502442 Medicine	OT	5	0
2502442 Medicine	OT	6	0

**Contact**

Name: Mercedes Campillo Grau  
Email: Mercedes.Campillo@uab.cat

**Use of Languages**

Principal working language: spanish (spa)  
Some groups entirely in English: No  
Some groups entirely in Catalan: No  
Some groups entirely in Spanish: No

**Teachers**

Leonardo Pardo Carrasco  
Ramón Guixa González  
Gianluigi Caltabiano  
Arnau Cordomi Montoya  
Angel Gonzalez

**Prerequisites**

Student taking this subject must have basic knowledge of English that allows him to navigate and understand the information contained in the databases and audiovisual material that he will have to consult in this language.

**Objectives and Contextualisation**

This course will illustrate how the discipline of bioinformatics provides an important bridge between cutting-edge science and the implementation of genomic medicine in clinical practice.

Students will be introduced to the basic concepts and tools of Bioinformatics focused on their future professional practice, through various activities to be carried out in the computer rooms.

The sessions aim to familiarize students with the use of the most used tools and online resources of the specialty.

Goals:

- Introduce the student in the genome / health world
- Initiate students in the use of tools, applications and types of data that are subject to analysis in clinical bioinformatics
- Introduce students in the concepts of medical informatics and precision medicine
- Introduce the student to ethical considerations in the use of genomic data

## Competences

### Medicine

- Be able to work in an international context.
- Critically assess and use clinical and biomedical information sources to obtain, organise, interpret and present information on science and health.
- Demonstrate basic research skills.
- Demonstrate understanding of the organisation and functions of the genome, the mechanisms of transmission and expression of genetic information and the molecular and cellular bases of genetic analysis.
- Demonstrate, in professional activity, a perspective that is critical, creative and research-oriented.
- Formulate hypotheses and compile and critically assess information for problem-solving, using the scientific method.
- Maintain and sharpen one's professional competence, in particular by independently learning new material and techniques and by focusing on quality.
- Organise and plan time and workload in professional activity.
- Use information and communication technologies in professional practice.

## Learning Outcomes

1. Be able to work in an international context.
2. Critique scientific papers on bioinformatics.
3. Demonstrate basic research skills.
4. Demonstrate, in professional activity, a perspective that is critical, creative and research-oriented.
5. Describe the diagnosis, prognosis, prevention and treatment for the most common genetic pathologies in the human population.
6. Formulate hypotheses and compile and critically assess information for problem-solving, using the scientific method.
7. Maintain and sharpen one's professional competence, in particular by independently learning new material and techniques and by focusing on quality.
8. Organise and plan time and workload in professional activity.
9. Use information and communication technologies in professional practice.

## Content

- Introduction to genomics and bioinformatics
  - Impact of the Human Genome Project on medicine
  - Genome sequencing: towards the diagnostic promise
  - Current status of genomic sequencing technologies and its clinical applications
- Bioinformatics and medical practice
  - What is OMIM and its usefulness in medical practice
  - Presentation of case studies as a common thread
- Ethical considerations in Bioinformatics and Genomics

## Methodology

The orientation of the subject is eminently practical and therefore the whole subject will be done directly in the computer rooms.

In many cases, learning includes the introduction and use of the main facilities offered by web applications and selected software.

The practices will be carried out individually or in pairs (depending on the number of students enrolled).

The student must perform a final consolidation work.

## Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
PRACTICAL SESSIONS (PLAB)	20	0.8	2, 4, 6
THEORY (TE)	5	0.2	2, 9
Type: Supervised			
Mentoring	5.25	0.21	2, 3, 6, 9
Type: Autonomous			
PERSONAL STUDY / READING OF ARTICLES / INTERESTING REPORTS	30	1.2	2, 3, 8, 1, 9
PREPARATION OF THE FINAL WORK	11	0.44	2, 3, 4, 8

## Assessment

The competences of the subject will be evaluated continuously, with assistance (40% of the grade), resolution of questionnaires (30% of the grade) and presentation of a final work (30% of the grade).

Students must obtain a minimum grade of 5 points to pass the course.

It will be considered that a student will obtain the qualification of "Not evaluable" if the qualification of the activities carried out does not allow him to reach a global score of 5 points in the event of having obtained the maximum grade in all of them.

Students who have not passed the course through continuous assessment may do additional work.

## Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Assistance and active involvement in the classes	40%	2	0.08	2, 5, 6, 8, 1, 9
Final work	30%	0	0	2, 3, 4, 6, 7, 8, 9
Resolution of questionnaires in the practical sessions	30%	1.75	0.07	7, 8

## **Bibliography**

### Reference bibliography

Attwood, T.K., Parry-Smith, D.J. Introduction to Bioinformatics. Pearson Education; 2002

Lesk, A. Introduction to Bioinformatics. Oxford University Press; 2005

Liang K-H. Bioinformatics for Biomedical Science and Clinical Applications. Woodhead Publishing; 2013

Sánchez Mendiola, M., Martínez Franco A.I. Biomedical Informatics. Elsevier; 2018

### Audiovisual resources:

Gattaca. Guión i Direcció: Andrew Niccol. Jersey Films / Columbia Pictures; 1997.

The DNA Journey. <https://youtu.be/2SB6ZaqEaLQ>

### Internet resources:

<https://www.ncbi.nlm.nih.gov/genome/gdv/>

<http://www.ncbi.nlm.nih.gov/clinvar/>

<http://www.genomesonline.org/index>

<https://ghr.nlm.nih.gov/>

<http://www.genecards.org>

<http://omim.org>

<http://www.genome.gov/GWASudies/>

<http://www.ebi.ac.uk/gwas/>