

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 |

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

Teachers

Leonardo Pardo Carrasco
Gianluigi Caltabiano
Arnau Cordomi Montoya
Angel Gonzalez

Prerequisites

To take this subject it is necessary to have basic knowledge of English to navigate and understand the information contained in the databases and audiovisual material to be consulted 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).

The minimum grade to pass the subject is 5 points.

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.

If the subject is not passed by continuous assessment, the recovery will consist of 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, Teresa K., Parry-Smith, David J. Introducción a la Bioinformática. Pearson Education; 2002.

Lesk, Arthur M. Introduction to Bioinformatics. Oxford University Press; 2014.

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/>