UAB
Universitat Autònoma de Barcelona

Medical Genomics and Bioinformatics

Code: 44344
ECTS Credits: 6

2025/2026

Degree	Type	Year
Advanced Genetics	OT	0

Contact

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Teachers

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Teaching groups languages

You can view this information at the end of this
document.

Prerequisites

A degree in the field of biosciences

Objectives and Contextualisation

Introduction to the applications of genomic medicine to the diagnosis, understanding and treatment of genetic diseases.

Introduction to the bioinformatic analysis of genetic variants, data bases, filtering of variants

Genetic councelling

Genetically based advanced therapies for the treatment of genetic disorders

Medical genetics and dysmorphology

Competences

- Demonstrate a mastery of genetic analysis as a transversal tool applicable to any field of genetics.
- Demonstrate responsibility in management of information and knowledge.
- Design and apply scientific methodology in resolving problems.
- Identify and propose scientific solution for problems related to genetic research at both molecular and organism levels and demonstrate an understanding of the complexity of living beings.
- 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.
- Student should possess an ability to learn that enables them to continue studying in a manner which is largely self-supervised or independent.
- Students should know how to apply the knowledge they acquire and be capable of solving problems in new or little-known areas within broader contexts (or multidisciplinary contexts) related to their area of study.
- Use and manage bibliographical information and other resources related to genetics and related fields.
- Use scientific terminology to argue the results of the research and show how to communicate in spoken and written English in an international setting.

Learning Outcomes

1. Apply bibliographical information about rules and legislation in risk assessment.
2. Demonstrate responsibility in the management of information and knowledge and in the direction of groups and/or projects in multidisciplinary teams.
3. Display knowledge of genetic analysis applied to the genomics of clinical cases.
4. Identify and compare the different methodologies of molecular analysis of genetic variability and medical genomics.
5. Identify suitable bioinformatic methodologies for genomic analysis applied to personalised medicine.
6. Preparation and presentation of seminars.
7. Solve practical problems in medical genomics by applying knowledge of bioinformatic analysis of the genoma.
8. Student should possess an ability to learn that enables them to continue studying in a manner which is largely self-supervised or independent.
9. Use scientific terminology to argue the results of the research and show how to communicate in spoken and written English in an international setting.
10. Write a report that considers the use of the methodology used in the module to resolve a specific problem.
11. Write critical summaries about the taught seminars.

Content

Introduction to the applications of genomic medicine to the diagnosis, understanding and treatment of genetic diseases. Introduction to the bioinformatic analysis of genetic variants, data bases, filtering of variants . Introcution to genetic councelling. Genetically based advanced therapies for the treatment of genetic diseases. Medical genetics and dysmorphology

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Bioinformatics in informatics classroom	25	1	1, 7, 2, 3, 11, 10, 4, 5, 6, 8, 9

Theoretica teaching	25	1	1, 7, 2, 3, 11, 10, 4, 5, 6, 8, 9
Type: Supervised			
Bioinformatic analysis	50	2	7, 5
Type: Autonomous			
Written report	40	1.6	7, 3, 11, 4

theoretical classes

Teaching based on problem solving
Classes in the bioinformatics classroom

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

Continous Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Exam of the Bioinformatics Part	40%	3	0.12	7, 3, 4, 5
exam theory	40%	4	0.16	1, 7, 2, 3, 11, 10, 4, 5, 6, 8, 9
Written report	20%	3	0.12	3, 11, 10, 5, 9

Exam of the theorethical teaching, problem solving at the bioinformatica classroom and written report

For the written work part of this subject, the use of Artificial Intelligence (AI) technologies is permitted exclusively in support tasks, such as bibliographic or information searches, text correction or translations. The student must clearly identify which parts have been generated with this technology, specify the tools used and include a critical reflection on how these have influenced the process and the final result of the activity. The lack of transparency in the use of AI in this assessable activity will be considered a lack of academic honesty and may lead to a partial or total penalty in the grade of the activity, or greater sanctions in serious cases.

Bibliography

to be showns during the teaching sessions

Software

Will be given in the PC classroom

Groups and Languages

Please note that this information is provisional until 30 November 2025. You can check it through this [link](#). To consult the language you will need to enter the CODE of the subject.

Name	Group	Language	Semester	Turn
(PLABm) Practical laboratories (master)	1	English	first semester	afternoon
(TEm) Theory (master)	1	English	first semester	morning-mixed