UAB
Universitat Autònoma de Barcelona

2023/2024

Medical Genomics and Bioinformatics

Code: 44344
ECTS Credits: 6

Degree	Type	Year	Semester
4313802 Advanced Genetics	OT	0	1

Contact

Name: Jordi Surralles Calonge

Email: jordi.surralles@uab.cat

Teaching groups languages

You can check it through this link. To consult the language you will need to enter the CODE of the subject. Please note that this information is provisional until 30 November 2023.

Teachers

Massimo Bogliolo

Lidia Gonzalez Quereda

Adriana Lasa Laborde

Benjamin Rodriguez Santiago

External teachers

Clara Serra

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

1

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

Methodology

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.

Activities

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

Assessment

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

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Bioinformatics problem resolution at the informatics classroom	25%	3	0.12	7, 3, 4, 5
Written report	25%	3	0.12	3, 11, 10, 5, 9
exam	50%	4	0.16	1, 7, 2, 3, 11, 10, 4, 5, 6, 8, 9

Bibliography

to be showns during the teaching sessions

Software

Will be given in the PC classroom