Genomics			2015/2016
Code: 42399 ECTS Credits: 12			
Degree	Туре	Year	Semester
4313473 Bioinformatics	ОТ	0	1

# Contact

# Use of languages

Name: Antoni Barbadilla PradosPrincipal working language: english (eng)Email: Antonio.Barbadilla@uab.catTeachersTeachersMario Cáceres AguilarJuan Ramón González RuízSònia Casillas ViladerramsRaquel Egea SánchezImage: english (eng)

### **External teachers**

Miguel Pérez-Enciso Miquel Ràmia Yolanda Guillén

# Prerequisites

To carry out this module is necessary to have passed previously both compulsory modules: *Programming in Bioinformatics* and *Core Bioinformatics*. Basic notions in genetics are also needed.

It is recommended you have a Level B2 of English or equivalent.

# **Objectives and Contextualisation**

The technological capacity to generate massive genomic data grows at a relentless pace without parallel growth of the bioinformatics expertise to deal with human, animal, microorganism and plant genomes. The purpose of this module is to provide the knowledge and technical skills which are required to successfully meet the current challenges of genomic analysis.

# Skills

- Analyse and interpret data deriving from omic technology using biocomputing methods .
- Communicate research results clearly and effectively in English.
- Design and apply scientific methodology in resolving problems.
- Identify the biocomputing needs of research centres and companies in the biotechnology and biomedicine sectors.

- 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.
- Propose biocomputing solutions for problems deriving from omic research.
- Propose innovative and creative solutions in the field of study
- Understand the molecular bases and most common standard experimental techniques in omic research (genomics, transcriptomics, proteomics, metabolomics, interactomics, etc.)
- Use and manage bibliographical information and computer resources in the area of study
- Use operating systems, programs and tools in common use in biocomputing and be able to manage high performance computing platforms, programming languages and biocomputing analysis.

### Learning outcomes

- 1. Communicate research results clearly and effectively in English.
- 2. Describe and apply the tools of assembly, annotation, storage, display and analysis of the variation in genomes.
- 3. Describe the operation, characteristics and limitations of first, second and third generation sequencing techniques.
- 4. Design and apply scientific methodology in resolving problems.
- Design and interpret studies of association between genetic polymorphisms and phenotype for identifying genetic variants affecting phenotype characters, including those associated pathologies and that confer susceptibility to human diseases or other species of interest.
- 6. Enumerate and describe the content of databases of relevant information for the different areas of genomics and carry out advanced searches.
- 7. Establish the corresponding relationships between nucleotide sequencing, structure and gene function using sources of biological data and the bases of biocomputing analysis.
- 8. Identify and characterize sources and display formats genomes, along with your notes and information about genetic variation, disease association and gene expression.
- 9. Integrating genomic data in silico reconstruction of cells and organisms (systems biology, synthetic biology).
- 10. 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.
- 11. Propose innovative and creative solutions in the field of study
- 12. Recognise the strategic importance of genetic advances in the area of human health, especially the genomic applications in personalised medicine and pharmacogenomics.
- 13. Use and manage bibliographical information and computer resources in the area of study
- 14. Use the latest algorithms of alignment of sequences and generation of evolutive trees as well as sequencing methods and gene prediction.

### Content

Lesson 1. Genome sequencing projects

### Lesson 2. Next Generation Sequencing (NGS)

Lesson 3. Primary NGS data analysis

### Lesson 4. Making sense of genome data

- 4.1 Genome assembly
- 4.2 Genome annotation
- 4.3 Functional analysis

### Lesson 5. Genome browsers

### Lesson 6. Genome variation

- 6.1 Theory and data
- 6.2 Nucleotide variation
- 6.3 Structural variation

#### Lesson 7. Association and GWA studies

#### Lesson 8. Expression analysis

- 8.1 Microarrays
- 8.2 RNAseq

**Lesson 9. Systems Genetics** 

#### Lesson 10. Applied genomics: solving problems and real cases

**Student Seminars' Session** 

### Methodology

The methodology will combine master classes, solving practical problems and real cases, work in the computing lab, performing individual and team works, reading articles and independent study student. It will use the virtual platform and asked for papers related to the thematic blocks.

### Activities

Title	Hours	ECTS	Learning outcomes
Type: Directed			
Seminars	4	0.16	
Solving problems in class and work in the computing lab	28	1.12	
Theoretical classes	37	1.48	
Type: Supervised			
Performing individual and team works	120	4.8	
Type: Autonomous			
Regular study	107	4.28	

### Evaluation

- Work done and presented by the student (student's portfolio) (60%).
- Individual theoretical and practical tests (40%)

# **Evaluation activities**

Title	Weighting	Hours	ECTS	Learning outcomes
Individual theoretical and practical test	40%	4	0.16	2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14

Student's portfolio	60%	0	0	1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14

# Bibliography

### **Basic references**

- Gibson, G. i S. V. Muse, 2009. A Primer of Genome Science. Sinauer, Massachusetts. 3rd edition.
- Barnes, M. 2007. Bioinformatics for geneticists (2nd Ed.) Wiley.
- Brown, T. A. 2007. Genomes. 3a ediciò. Garland Science.
- Pevnser, J. 2009. Bioinformatics and Functional Genomics (2nd edition). Wiley-Blackwell.
- Samuelsson, T. 2012. Genomics and Bioinformatics: An Introduction to Programming Tools for Life Scientists. Cambridge University Press.

### **Recomended Websites**

- Course: Gurrent topics in Genome Analysis 2014. NHGRI (http://www.genome.gov/12514288)
- National Human Genome Research Institute (USA) (http://www.genome.gov/)
- Genomic careers (http://www.genome.gov/genomicCareers/video\_find.cfm)
- Genome online databases (GOLD) (https://gold.jgi-psf.org/index)
- Map Viewer NCBI (http://www.ncbi.nlm.nih.gov/projects/mapview/)
- Ensembl genome browser (http://www.ensembl.org/index.html)
- UCSC genome browser (http://genome.ucsc.edu/)
- Genome size databases (http://www.genomesize.com/)