

Degree	Type	Year
2500890 Genetics	OB	3

Contact

Name: Sonia Casillas Viladerrams

Email: sonia.casillas@uab.cat

Teachers

Javier Garcia Pardo

Teaching groups languages

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Prerequisites

Although there are no oficial requirements, students are assumed knowledge of Biochemistry and Molecular Biology, Genetics, Microbiology, Cellular Biology, Methods of recombinent DNA and Statistics.

For some activities a basic level of english is necessary.

Objectives and Contextualisation

Life as we know it is specified by the genomes of the thousands of organisms with which we share the planet. Each of these genomes contains the biological information needed to build and keep alive a specimen of the corresponding organism. The development of techniques that allow us to read the sequence of these genomes has opened up the possibility of getting to know us better and starting to look for the answer to questions such as: What are we? How have we become what we are? What do we share with other humans and what makes us different from other members of our species? What similarities and differences do we have with our closest relatives, the chimpanzees? Or with other species of primates? Mammals? Vertebrates? Eukaryotes? With all other living things? Or even, what the future holds for us?

Genomics is the science that studies the structure, content, and evolution of genomes. It is a relatively new science (born in 1995 with the sequencing of the first bacterial genomes) and has developed explosively in the recent years. The development of automatic nucleic acid sequencing methods has been key. In 2001, the first draft of the sequence of the human genome was presented. It represented a historic landmark which opened the doors to studies about comparative genomics and evolution of the human species, the biological components of human kind, genotype-phenotype association studies for the discovery of disease-related genes or regions of DNA, and so on.

The improvement in omic techniques has not only resulted in an explosive growth in the quantity and quality of genomic data, but has also opened up the possibility of studying other aspects of molecular biology such as massive analysis of the expression of genes and genomes (Transcriptomics and Functional Genomics), the identification and structural and functional analysis of proteins (Proteomics) and their interactions (and with other biomolecules) and the formation of complexes (Interactomics). In conjunction with the identification and quantification of all the metabolites present in a sample of the organism (Metabolomics), this knowledge gives rise to the bases to integrate all the data and reach a global description of the biology of the cell (Systems Biology).

The main objectives of this course are: (i) to understand the structure, diversity and complexity of genomes, transcriptomes and proteomes; (ii) to recognize the functional, historical, and evolutionary nature of genetic information, as well as the nature, meaning, and consequences of intra-specific and inter-specific variability; and (iii) to learn experimental and computational methods as well as potential applications of genomics, transcriptomics, and proteomics.

Competences

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
- Apply knowledge of theory to practice.
- Apply scientific method to problem solving.
- Be able to analyse and synthesise.
- Describe and identify the structural and functional characteristics of nucleic acids and proteins including their different organisational levels.
- Describe the organisation, evolution, inter-individual variation and expression of the human genome.
- Design and interpret studies associating genetic polymorphisms and phenotypical characters to identify genetic variants that affect the phenotype, including those associated to pathologies and those that confer susceptibility to human illnesses or those of other species of interest.
- Design experiments and interpret the results.
- Develop self-directed learning.
- Know and apply the omic' tools of genomics, transcriptomics and proteomics.
- Know and interpret the metabolic and physiological bases of organisms.
- Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
- Perceive the strategic, industrial and economic importance of genetics and genomics to life sciences, health and society.
- Reason critically.
- Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
- Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
- Use and manage bibliographic information or computer or Internet resources in the field of study, in one's own languages and in English.

Learning Outcomes

1. Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
2. Apply knowledge of theory to practice.
3. Apply scientific method to problem solving.
4. Be able to analyse and synthesise.
5. Defend the relevance of progress in the generation and interpretation of data on a genomic scale for our understanding and technological manipulation of organisms.
6. Describe and apply the methods for the analysis of proteomes, of genomics and of functional proteomics.

7. Design experiments and interpret the results.
8. Develop self-directed learning.
9. Explain and apply the methods for the analysis and annotation of genomes.
10. Explain how the joint use of high performance technologies to study genetic variation, bioinformatics resources and statistical methods make it possible to exhaustively catalogue the genetic variants that affect the phenotype.
11. Explain the basics of metabology/metabolics and their methods.
12. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
13. Reason critically.
14. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
15. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
16. Use and manage bibliographic information or computer or Internet resources in the field of study, in one's own languages and in English.
17. Use bioinformatics techniques and tools to describe and analyse the human genome.
18. Use the techniques, tools and methodologies used to describe, analyse and interpret the enormous amounts of data produced by high performance technologies.

Content

PART I. GENOMICS

Topic 1. Genomes: structure and function

Topic 2. Genome sequencing strategies

Topic 3. Sequencing, assembly and annotation of genomes

Topic 4. Study of gene expression: transcriptomics

Topic 5. Genome evolution: comparative genomics

Topic 6. Genomic and paleogenomic variation

PART II. PROTEOMICS AND INTERACTOMICS

Topic 7. Proteomics: introduction and basic concepts

Topic 8. Proteome diversity

Topic 9. Basic techniques of proteome analysis

Topic 10. Quantitative proteomics

Topic 11. Structural and functional proteomics

Topic 12. Interactomics

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
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Type: Directed

Seminars and problem solving classes	15	0.6	2, 3, 4, 5, 6, 7, 9, 10, 11, 13, 16, 17, 18
Theory	30	1.2	2, 3, 4, 5, 6, 7, 9, 10, 11, 13, 16, 17, 18
Type: Supervised			
Tutoring	3	0.12	2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 13, 16, 17, 18
Type: Autonomous			
Problem solving, activities and recommended readings	60	2.4	2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 13, 16, 17, 18
Study	42	1.68	2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 13, 16, 17, 18

The subject consists of theoretical classes, seminars for the resolution of practical cases and problems, and tutoring sessions. Here we describe the organization and the teaching methodology that will be followed in these three types of training activities.

Theory classes:

The content of the theory program will be taught mainly by the professor in the form of master classes with audiovisual support. Presentations used in class by the professor will be previously available on the Moodle classroom of the subject. It is recommended that students have this material in class, to use it as a support when taking notes. It is recommended that students regularly consult the books recommended in the bibliography section of this teaching guide, as well as the readings proposed in the Moodle classroom, in order to consolidate and clarify, if necessary, the contents explained in class.

Seminars and problem solving classes:

The mission of the seminars and problem solving classes is to bridge between master classes and practical work, promoting active learning to develop the capacity for analysis and synthesis, critical reasoning and problem solving ability. The seminars are sessions with a small number of students (maximum 30 students) where to deepen or complete the knowledge presented in the master classes by solving problems and discussing case studies. Students will receive periodic recommended readings, problems and cases to solve, web addresses to navigate, etc.

Tutoring sessions:

There will be up to 3 tutoring sessions with the professor. In these sessions, the doubts that the students raise about the theoretical program topics will be resolved to facilitate the understanding of specific aspects of the subject.

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

Continuous Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
ADAPT YOURSELF (integrative problem of the whole subject)	10%	0	0	1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18

PHYLOGENOME (integrative work of Genomics)	10%	0	0	1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18
Partial exam 1 (Genomics)	25%	0	0	2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 13, 16, 17, 18
Partial exam 2 (Proteomics/Interactomics)	35%	0	0	2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 13, 16, 17, 18
Seminars of Proteomics/Interactomics	10%	0	0	2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 13, 16, 17, 18
Weekly problems of Genomics	10%	0	0	2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 13, 16, 17, 18

The evaluation system is organized in six main activities. There will also be a recuperation exam and an optional activity to improve the final grade. The details of the activities are:

Main evaluation activities

(A) Partial exams. Overall weight 60%

- Partial exam 1 (Genomics). Weight 25%
- Partial exam 2 (Proteomics/Interactomics). Weight 35%

Partial exams are combined tests that can consist of multiple-choice questions or short answer questions or problems. These tests will be eliminatory of contents.

There will be a first partial exam corresponding to the part of Genomics with a weight of 25% on the final grade of the subject, and a second partial exam corresponding to the part of Proteomics/Interactomics with a weight of 35% on the final grade of the subject.

To pass these two evaluations you have to reach a minimum score of 4.0 in each of them.

(B) Continuous assessment. Overall Weight 40%

Throughout the course, professors will present problems or seminars related to the taught subject that the students must solve in the form of evaluations, deliveries or active participation in the seminars. There will be four different typologies:

- Weekly Problems of Genomics. Weight 10%

The continued nature of this evaluation means that the subject can not be evaluated unless there is a minimum participation in 50% of the activities proposed.

- PHYLOGENOME (integrative work of Genomics). Weight 10%

The work will contain issues related to the different topics of the Genomics part (<https://phylogenome.omicsuab.org>). It will be done in groups of 3-5 students. Professors will supervise the work and evaluate progress on a weekly basis and at the end of the subject.

Students will deliver the works following the guidelines for content, presentation and deadlines.

This activity is mandatory and non-participation will entail non-evaluation of the subject.

- Seminars of Proteomics/Interactomics. Weight 10%

The continued nature of this evaluation means that the subject can not be evaluated unless there is a minimum participation in 50% of the activities proposed.

- ADAPT YOURSELF (integrative problem of the whole subject). Weight 10%

This activity is linked to the ADAPT YOURSELF teaching innovation project and gives the option to participate in a learning and service experience in the second semester, recognized with 2 ECTS (<https://adaptat.omicsuab.org>).

The problem will contain issues related to the different topics of the two parts of the subject (Genomics and Proteomics/Interactomics).

Students will deliver the problem following the guidelines for content, presentation and deadlines.

This activity is mandatory and non-participation will entail non-evaluation of the subject.

Retake exam

To be eligible for the retake process, the student should have been previously evaluated in a set of activities equaling at least two thirds of the final score of the course.

The evaluations 1 and/or 2 can be retaken individually and the grade, if ≥ 4 , will make an average with those passed in the partial exams. In the event that the grade of the retake exam does not reach a minimum of 4.0, it cannot make an average and the student does not pass the subject.

Continuous assessment (weekly problems of genomics, integrative work of genomics, seminars of proteomics/interactomics and integrative problem of proteomics/interactomics), due to its continued nature, cannot be retaken.

Improvement of the final qualification

Students who have passed the evaluations 1 and 2 want to improve their final qualification, may opt for a final test. This test will include all the subject contents. It is not possible to improve the grade through works or other types of activities.

The degree of difficulty of this test will correspond to the objective of the test and, therefore, may be higher than the partial evaluations.

The student presented in this test waives the qualifications of the partial evaluations 1 and 2. Therefore, the grade of this improvement test will be the one that will prevail in the final grade even though it is lower than that obtained in the partial evaluations.

Formula of weighting of the final grade

Final grade = [(Partial exam 1 x 0.25) + (Partial exam 2 x 0.35) + (Weekly Problems of Genomics x 0.1) + (Integrative work of Genomics x 0.1) + (Seminars of Proteomics/Interactomics x 0.1) + (Integrative problem of the subject x 0.1)]

Passed

The subject is considered passed if the final grade is ≥ 5 .

Not evaluable

The student will be graded as "No Evaluable" if the weight of all conducted evaluation activities is less than 67% of the final score.

Unique assessment

The unique assessment consists of a single synthesis test in which the contents of the entire theory program of the subject will be evaluated in the form of a theoretical-practical test. The test will consist of multiple-choice questions, written answers and problem solving. The grade obtained in this synthesis test will account for 60% of the final grade of the subject (25% for the Genomics part, and 35% for the Proteomics/Interactomics part).

To pass this test you must achieve a minimum grade of 4.0 in each of the two parts of the test (Genomics and Proteomics/Interactomics). The single assessment test will coincide with the same date fixed in the calendar for the last partial assessment test and the same recovery system will be applied as for the conventional assessment.

The assessment of the different Continuous Assessment activities (Weekly problems of Genomics, Genome Project (integrative work of Genomics), Proteomics/Interactomics Seminars, ADAPTA'T (integrative problem of the whole subject)) will follow the same process of conventional assessment. The grade obtained will account for 40% of the final grade of the subject (10% for each of the four activities). Students who take the single assessment can submit these four pieces of evidence together on the same day as the one set for the synthesis test.

Students taking the single assessment must take the Proteomics/Interactomics Seminars in face-to-face sessions and under the same conditions as in the conventional assessment.

Bibliography

The Moodle classroom of the subject (<https://omics-elearning.omicsuab.org>) is the didactic resource of reference of the course. There, all teaching materials are provided to students, including PowerPoint presentations, recommended readings, videos, weekly problem questionnaires, project information, and more. It is also used to manage work groups, submissions and grades, publish news or notices, collect survey results and facilitate teacher-student communication.

Students should try to consult as many bibliographic sources and textbooks as possible, which will provide them with enough material to be able to confront, expand and deepen the different concepts that are worked on throughout the course. In addition, in the field of omics, techniques and knowledge are advancing at a frantic pace and only publications in scientific journals manage to keep up. Thus, while some teaching units are well covered in textbooks, others will need to be complemented by the latest publications in the field. For this reason, the material that will be provided throughout the course will provide references to the sources from which it was extracted.

Recommended textbooks:

Brown TA. 2023. Genomes 5. Garland Science, New York, USA. Fifth edition. [topographic 575.113 Bro]
A very complete and easy-to-follow textbook, which closely follows the structure of the GPI subject: first the techniques are presented, then the anatomy of the genome, followed by the function of the genome and finally its evolution. The figures and diagrams stand out, so in many cases they have been used to illustrate the materials of the subject. It also includes a set of short answer questions and discussion problems at the end of each chapter.

Lesk AM. 2017. Introduction to genomics. Oxford University Press, Oxford, UK. Third edition. [topographic 577.113 Les]
A very complete and up-to-date textbook, with a very attractive style of writing that provides a fascinating insight into all that can be revealed about the study of omics. Accompanied by a good number of exercises and problems in each topic, the online resource center stands out, which includes web-based problems, tips and answers to the problems and exercises at the end of each chapter, a guided tour of websites and large molecular databases, links to complementary readings, discussions, and interactive figures.

Gibson G and Muse SV. 2009. A primer of genome science. Sinauer Associates, Inc., Sunderland, MA, USA. Third edition. [topographic 575.113 Gib]
Brief and affordable introduction to genomics, proteomics, metabolomics and interactomics. Although it has become a bit outdated, the figures and diagrams, the boxes with explanations of bioinformatics methods and the online resource center stand out, which includes exercises for each chapter with links and downloadable files, as well as links to all websites referenced throughout the book.

Twyman RM. 2014. Principles of proteomics. Garland Science, New York, USA. Second edition. [Topographic 577.112 Twy]

It provides a concise and easy-to-use introduction to the various technologies used for large-scale protein analysis, as well as their applications and impact in areas such as drug discovery and agriculture. The book includes examples, case studies, and complementary readings in each chapter.

Lovric J. 2011. *Introducing proteomics*. John Wiley & Sons, Oxford, UK. [Ebook]

It provides a concise and consistent view of all aspects of current proteomics technology. It covers the analysis of peptide and protein separation technologies using mass spectrometry strategies and includes numerous examples and explanations of why some strategies are better than others for certain applications.

Mirzaei H I Martin C. 2016. *Modern proteomics: Sample preparation, analysis and practical applications*. Springer, Berlin, Germany. [Ebook]

Proteomics Reference Manual, which describes experimental design and execution. It also shows a wealth of examples of what can be achieved through proteomics techniques, with a particular focus on technical details.

Software

Databases:

- Animal Genome Size Database, <http://www.genomesize.com/>
- Ensembl Genome Browser, <http://www.ensembl.org>
- Flybase JBrowse, <http://flybase.org/jbrowse/?data=data/json/dmel>
- GOLD Genomes Online Database, <https://gold.jgi.doe.gov/>
- NCBI Genome, <http://www.ncbi.nlm.nih.gov/genome>
- PopHuman Genome Browser, <https://pophuman.uab.cat>
- PopHumanScan, <https://pophumanscan.uab.cat>
- PopHumanVar, <https://pophumanvar.uab.cat>
- UCSC Genome Browser, <http://genome.ucsc.edu/cgi-bin/hgGateway>
- Genome Reference Consortium, <https://www.ncbi.nlm.nih.gov/grc/human>
- RepeatMasker, <http://www.repeatmasker.org/genomicDatasets/RMGenomicDatasets.html>
- BioGPS, <http://biogps.org/>
- ENCODE, <http://genome.ucsc.edu/ENCODE/>
- Expression Atlas, <https://www.ebi.ac.uk/gxa/home>
- GENCODE Human, <https://www.gencodegenes.org/human/>
- InterMine, <http://intermine.org/>
- Synteny Portal, http://bioinfo.konkuk.ac.kr/synteny_portal/
- VISTA Genome Browser, <http://genome.lbl.gov/vista/index.shtml>
- International Cancer Genome Consortium, <https://icgc.org/>
- Open Targets, <https://www.targetvalidation.org/>
- The Cancer Genome Atlas, <https://www.cancer.gov/about-nci/organization/ccg/research/structural-genomics/tcga>
- Human protein Atlas, <https://www.proteinatlas.org/>
- Human proteome map, <http://www.humanproteomemap.org/>
- Uniprot, <https://www.uniprot.org/>
- SWISS-2D, <https://world-2dpage.expasy.org/swiss-2dpage/>
- Phosphosite, <http://www.phosphosite.org/>
- Peptide Atlas, <http://www.peptideatlas.org/>
- SRMATlas, <http://www.srmatlas.org/>
- Gene Ontology, <http://geneontology.org/>
- String, <https://string-db.org/>

Analysis tools:

- ScanProsite, <http://prosite.expasy.org/scanprosite/>
- Transeq, http://www.ebi.ac.uk/Tools/st/emboss_transeq/
- GeneID, <http://genome.crg.es/geneid.html>
- RevTrans, <http://www.cbs.dtu.dk/services/RevTrans-2.0/web/>

- MASCOT <http://www.matrixscience.com/>
- ProtParam <https://web.expasy.org/protParam/>
- SwissModel <https://swissmodel.expasy.org/>
- Provean <http://provean.jcvi.org/index.php>
- INPS-server <https://inpsmd.biocomp.unibo.it/inpsSuite>

Language list

Name	Group	Language	Semester	Turn
(PAUL) Classroom practices	631	Catalan/Spanish	first semester	morning-mixed
(PAUL) Classroom practices	632	Catalan/Spanish	first semester	morning-mixed
(SEM) Seminars	631	Catalan/Spanish	first semester	morning-mixed
(SEM) Seminars	632	Catalan/Spanish	first semester	morning-mixed
(TE) Theory	63	Catalan/Spanish	first semester	morning-mixed