Universitat Autònoma de Barcelona

2022/2023

Laboratory VI

Code: 101942
ECTS Credits: 3

Degree	Type	Year	Semester
2500890 Genetics	OB	3	2

Contact

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Use of Languages

Principal working language: catalan (cat)
Some groups entirely in English: No
Some groups entirely in Catalan: No
Some groups entirely in Spanish: No

Prerequisites

- It is mandatory to have taken -o being currently taking- the theoretical subjects related to the experimental work developed here.
- Biosecurity and security tests at 'campus virtual' need to be passed. The student must prove knowledge and acceptance of the Bioscience laboratory guidelines.
- It is necessary to go through the theoretical content of each module before the day of the practical classes.
- Wearing a lab coat is mandatory. It is not possible to enter to the lab without a lab coat.
- Attendance is mandatory.
- Students should come to the class following the assigned schedule. Changes in the original schedule need to be approved by the corresponding professor and in all cases before the starting of the classes.

Objectives and Contextualisation

The Integrated Laboratory VI is the sixth course in a set of 6 which are distributed along 6 semesters of the first three courses of the degree of genetics. These subjects aim to give a solid foundation of experimental procedures, techniques and skills of genetics and related sciences. The practical work help to reinforce the theoretical concepts acquired in the theory, and allow us to understand the essential dialogue between theory and experimentation that have given rise to the body of knowledge that constitutes the science of genetics.

The Integrated Laboratory VI has as objectives the acquisition of experimental skills in 3 specific modules of content:

- Molecular genetic diagnostic
- Evolution
- Systems Biology

Module of Molecular genetic diagnosis

The main objective of the module is to know and apply some of the basic techniques of molecular genetics in the diagnosis of diseases.

Module of Evolution

The main objective of the Evolution module is to help the student to learn to apply the comparative method in evolutionary inference by using molecular sequences. The module includes the processes of hypothesis formulation and testing, obtention of sequence data for their alignment, modeling of the process of evolutionary change of the sequences, and application of methods of i) phylogenetic reconstruction, ii) characterization of the evolution of gene families, and iii) Identification of footprints of molecular adaptation. In addition, a perspective on the levels and structure of genetic variation in *Homo sapiens* will be provided by comparison with what is observed in other closely related living and extinct species. Through these practices the student will acquire skills in the application of analytical techniques to biological and molecular data.

Module of Systems biology

The main objective of the module is to learn how to use the right software in systems biology and better understand the behavior of biological systems. To achieve this the student will conduct simulation exercises with computer to see how the behaviorof a biological system cannot be predicted from the behavior of its isolated components.

Competences

- Appreciate the importance of quality and a job well done.
- Describe and identify the structural and functional characteristics of nucleic acids and proteins including their different organisational levels.
- Design and execute complete protocols of the standard techniques that form part of molecular genetics instruments: purification, amplification and sequencing of genomic DNA from biological sources, genetic engineering in microorganisms, plants and animals.
- 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.
- Know and apply the omic' tools of genomics, transcriptomics and proteomics.
- Perform genetic diagnoses and assessments and consider the ethical and legal dilemmas.
- Understand and describe the structure, morphology and dynamics of the eukaryotic chromosome during the cell cycle and meiosis.
- Use and interpret data sources on the genomes and macromolecules of any species and understand the basics of bioinformatics analysis to establish the corresponding relations between structure, function and evolution.

Learning Outcomes

1. Apply biochemical and genetic engineering techniques to identify and characterise nucleic acids and proteins.
2. Apply global analysis software to the integration, representation and modelling of networks for interconverting biological information.
3. Apply techniques for the genetic engineering of microorganisms, plants and animals in specific genetic, medical and agricultural problems
4. Appreciate the importance of quality and a job well done.
5. Build and use gene libraries.
6. Describe high performance data production systems by visiting the UAB's microarray and genotyping centres and services.
7. Detect polymorphisms associated to illnesses and other phenotypical characters of interest in humans and livestock.
8. Detect specific molecules of DNA, RNA and proteins using hybridisation probes.
9. Enumerate and describe the contents of databases containing information of relevance to the different fields of genetics and perform advanced research.
10. Perform genetic diagnoses and assessments on the basis of molecular analysis of diagnostic mutations.
11. Perform tests to identify individuals or specimens using their genetic fingerprint.

12. Prepare, observe and recognise polytene chromosomes of Drosophila.
13. Provide genetic warnings by detecting markers that are associated to illnesses.
14. Use PCR for genetic diagnosis.

Content

Module of Molecular genetic diagnostic

Use of different basic molecular genetic techniques applied to specific diagnostic cases: PCR and sequencing, MLPA and Western blot.

Module of Evolution

a) Alignment: obtention, reliability and interpretation.

b) Modeling of molecular evolution: problem of multiple substitutions and selection of optimal models of sequence evolution.

c) Molecular phylogenetic trees: obtention, reliability and interpretation.

d) Distribution of character states. Evolution of gene families.

e) Molecular dating. Human genetic diversity

Module of systems biology

The module includes practical exercises corresponding to the theory of the subject ' systems biology' and will consist in the simulation of systems for example corresponding to genetic, metabolic networks, signal transmission or of more complex systems, developed by the student with the recommended software or downloaded from databases of models according to the case.

In general the practices include exercises for the use of software and databases specific to each of the topics. These exercises will allow.

Methodology

The subject is taught in small groups of students (maximum 20 per session) in the laboratory or computer rooms. Students have a manual or practice guide for each Module. It is necessary to read the corresponding part of each session carefully before starting the practice to obtain the maximum advantage. Students will have to attend the assigned group of internships obligatorily. Only occasional changes will be accepted as long as they are balanced (a student from a group for a student from another group). If a student has not been able to perform a practice session with his group, he can retrieve it by attending another group, as long as the group in question has free spaces.

Molecular genetic diagnosis

The students will have a detailed guideine of the practices with the protocols and the detailed information to be able to carry out them efficiently with the support of the teacher. The guideline will be available on the Virtual Campus of the subject. The students will not only work their data, but will analyze and interpret the set of results obtained.

Evolution

The Practicum will take place in computer rooms using several applications. Data obtained from scientific publications will be used. Different concepts and hypotheses about the evolutionary process will be illustrated through presentation of real problems. A guide to the Practicum will be available on the Virtual Campus of the subject.

Systems biology

Systems biology practices will be carried out in the computerrooms on days and hours defined in the teaching calendar.

For each practice, the student will find the practical guidelines on the virtual campus of the subject 'Systems Biology'.

The student will carry out the practice following the practice guideline and save the files generated in the folder of his personal record provided by the UAB.

When finishing the practice, the student will deliver, through the virtual campus, the files of the exercises as described in each practice.

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			
Evolution Module	15	0.6	7, 9, 12
Module Molecular Systems Biology	15	0.6	2, 9, 4
Module Molecular genetic diagnosis	16	0.64	1, 3, 5, 6, 8, 7, 10, 11, 14, 4
Type: Supervised			
Individual tutorials	1	0.04	
Type: Autonomous			
Lab booknote	3	0.12	
Study	23	0.92	

Assessment

Attendance to practical sessions is mandatory. Students missing more than 20% of programmed sessions will be graded as "No Avaluable".

Attendance to the practices is mandatory and therefore an absence without justification may entail the non-evaluation of one or more modules. Missing a session implies a reduction of the grade equal to% of this session in the whole module. Thus, in a module of 4 sessions, missing a day involved a reduction of 25% of the note in this module. Those students who can not attend the session of their group for just cause are exempt from this penalty. Health problems are deemed justified (the corresponding medical certificate must be carried out by the practice coordinator) or serious personal problems. In this case the practice will be recovered whenever possible.

Notwithstanding other disciplinary measures deemed appropriate, and in accordance with the current academic regulations, the irregularities committed by the student who can lead to a variation in the qualification of an act of self- evaluation. Therefore, copying or letting copy a practice or any other evaluation activity will

imply suspending it with a zero, and if it is necessary to pass it to pass it, the entire subject will be suspended. Qualified assessment activities will not be recovered in this way and by this procedure, and therefore the subject will be suspended directly without opportunity to recover it in the same academic year.

Module Molecular genetic diagnosis

This module will be evaluated by means of a final exam about the experimental procedure of the practices, that will take place the last day of the practices and will count 40% of the note. The other 60% of the mark will be evaluated by delivering a practical script. In the event that the student suspends the examination, the exam may be recovered, but the maximum grade that can be obtained in the recovery will be 5. The possibility of notifying notes is contemplated. The delivery note can not be recovered in any case.

Evolution Module

The module will be evaluated with a practical test of the contents included in the module. The final qualification of the module will take into account the attitude and work of the student in the classroom.

Systems Biology

Grading of the Systems Biology module will be calculated as follows:

First part of the grade (40%) will be obtained from practical sessions 1 to 4. It will be obtained from the responses to the proposed questionnaire as well as from the proposed exercises as described in each guide for the practical work.

The second part of the grade (60%) will be obtained from the 5^{th} practical class (last one) from the delivered simulator files and the answers provided to the proposed questions, delivered in the summary document for the 5^{th} practical class.

In case the weighted average obtained using the above procedure would not be equal or higher to 5, the student can take a remedial exam.

The final grade is the average of the notes of each module.

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Evolution Module	33%	0.7	0.03	9, 12, 4
Module Molecular biology of systems. Continuous evaluation of the results worked	33%	0.65	0.03	2, 9, 4
Module Molecular genetic diagnosis. Continuous evaluation of the results worked	33%	0.65	0.03	1, 3, 5, 6, 8, 7, 13, 10, 11, 14, 4

Bibliography

The scripts of the different practices will contain the specific bibliography of each one of them.

Software

- COPASI (http://copasi.org/)