Universitat Autònoma de Barcelona

2022/2023

Laboratory V

Code: 101943
ECTS Credits: 3

Degree	Type	Year	Semester
2500890 Genetics	OB	3	1

Contact

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Use of Languages

Principal working language: catalan (cat)
Some groups entirely in English: No
Some groups entirely in Catalan: Yes
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 V is the fifth 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 V has as objectives the acquisition of experimental skills in 3 specific modules of content:

- Genomics
- Human Genetics
- Quantitative genetics and improvement

Genomics

The main objective of Genomics module is to understand the process of Assembly, annotation and analysis of genomic sequences. Apart from learning to work with DNA sequences and proteins also will acquire knowledge about the structure and the characteristics of the various functional elements that can be found in a genome.

Human Genetics

The aim of the module of human genetics is know how to identify genetic mutations and polymorphisms related to the generation of diversity and the pathological processes. Through these practices, the student will acquire skills in the application of instrumental, molecular and analytical techniques.

Quantitative genetics and improvement

The aim of this module is to help you understand the principles of Quantitative genetics and its application in the selection, as well as the availability of tools for the identification of individual genes that determine the complex characters.

Competences

- Appreciate the importance of quality and a job well done.
- 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.
- Measure and interpret the genetic variation in and between populations from a clinical, conservational and evolutionary perspective, and from that of the genetic improvement of animals and plants.
- Perform genetic diagnoses and assessments and consider the ethical and legal dilemmas.
- 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. Analyse and interpret human genetic variation.
2. Apply and interpret simulation programs to improve livestock farming.
3. Apply global analysis software to the integration, representation and modelling of networks for interconverting biological information.
4. Apply techniques for the genetic engineering of microorganisms, plants and animals in specific genetic, medical and agricultural problems
5. Apply the basic common techniques of a human genetics laboratory.
6. Appreciate the importance of quality and a job well done.
7. Design the methodology, sampling and selection of genetic markers and the statistical analyses required in an association study.
8. Detect polymorphisms associated to illnesses and other phenotypical characters of interest in humans and livestock.
9. Determine the base sequence of a DNA segment.
10. Enumerate and describe the contents of databases containing information of relevance to the different fields of genetics and perform advanced research.
11. Perform genetic diagnoses and assessments on the basis of molecular analysis of diagnostic mutations.
12. Perform tests to identify individuals or specimens using their genetic fingerprint.
13. Predict the risk of the genetic transmission of illnesses or of other characters of interest by analysing the genealogies of humans and livestock.
14. Provide genetic warnings by detecting markers that are associated to illnesses.
15. Use databases on haplotype variation and genotype-phenotype association in humans and livestock.
16. Use statistical techniques and available software to perform association studies.

Content

Module of Genomics

The module is organized in 5 sessions of 3 hours each that will take place in the computer room. The work will consist of the Assembly, the annotation and analysis of a sequence. Based on initial data, the practice will be continuing along the 5 sessions so that each of them will be one more step in the process or will examine a different aspect of the secuence. The work will be distributed in the following way:

Session 1. Assembly

Session 2. Scaffolding

Session 3. gene annotation Ab initio and by homology

Session 4. Annotation of genes with RNA-seq

Session 5. Functional analysis and discussion

Module of Human Genetics

The module is organized in 4 sessions of 4 hours each that will be carried out in the laboratory. The students will go through three possible cases occurring in a genetics laboratory: case of prenatal diagnosis, case of leukemia, population screening for a mutation with possible applications (e.g. in the pharmacogenetics or Nutrigenetics). To respond to these three situations several techniques will be used, such as conventional cytogenetics, fluorescence in situ hybridization (FISH, polymerase chain reaction (PCR) and restriction fragment length polymorphisms (RFLPs).

Module of Quantitative genetics and improvement

GQM module is organized in 6 sessions that will take place in the computer room. The sessions will be synchronic with the theory classes (see calendar), so that the student may work and reflect the essential concepts and methodologies of the topic. The sessions are the following:

Session 1. Analysis of the genetic components of quantitative characters (2 h).

Session 2. Analysis of Association (Genome-wide association studies), and (3).

Session 3. Analysis of Association (Genome-wide association studies), II (2 h).

Session 4. Genetic assessment: animal model BLUP (2 h).

Session 5. The optimization selection and effects of selection on the genetic structure of populations (3 h).

Session 6. Simulation of the selection in the cow (3 h).

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.

Genomics Module

Self-guided learning guided by the teacher / a by the practitioner. Students will have to elaborate the data, interpret the results obtained and respond to the different questions raised in the practice guideline.

Human Genetic Module

The student will perform the experimental work independently following the practice guideline and with the help of the practice lecturer. After obtaining the results, these will have to be analyzed and interpreted. In the practicals guide there will be some questions to help with this analysis and interpretation of the results.

Quantitative Genetics Module
Each session begins with an introduction by the teacher and then the student must follow the practice guidelines and obtain their own results, always under the supervision of the teacher.

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			
Genomics Module	15	0.6	3, 9, 10, 6
Human Genetics Module	16	0.64	1, 5, 4, 8, 14, 11, 6
Quantitative Genetics Module.	15	0.6	1, 2, 8, 7, 13, 15, 16, 6
Type: Supervised			
Individual tutorials	1	0.04	
Type: Autonomous			
Laboratory notebook	5	0.2	
Study	23	0.92	

Assessment

Attendance 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 justify causes 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.

Genomics Module
The practices will be evaluated by delivering the results obtained during the practice that will have to be presented in a clear and understandable way.
The correction of the data obtained will be assessed. The attitude and work of the student in the classroom will also be taken into account.

Human Genetics Module
The practices will be evaluated by delivering the questions of the practice script and the results obtained during the practice. The interpretation that is made of the obtained data will be valued especially. The attitude and work of the student in the laboratory will also be taken into account.

Quantitative Genetics Module
It will be assessed by completing the completed practice script. The accuracy in the resolution of the questions raised will be taken into account, clarity in the presentation of comments and conclusions, and also the formal presentation. The student's attitude will also be evaluated during the practices.
To pass the course, it is necessary to first approve each module with a mark ≥ 5. Students who do not pass the different modules of the subject will be able to recover them on the scheduled date for the recovery evaluation of the subject.
The student who has not passed one of the modules after the recovery evaluation will not approve the subject. In spite of this, it will not be necessary for a repeating student to carry out the teaching activities or the evaluations of that module passed after the second enrollment.
Repeaters will only have to be evaluated for thespecific module that has not been exceeded. This exemption will be maintained for a period of three additional license plates.
The final grade is the average of the notes of each module. Not evaluable The "Non-Valuable" qualification will be obtained when the number of assessment activities performed is less than 50% of the programmed ones.

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 or module.
Thus, the student will be graded as "No Avaluable" if the weighthin of all conducted evaluation activities is less than 67% of the final score.

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

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Genomics module. Continuous evaluation of the results worked	33.3%	0	0	3, 9, 10
Human Genetic Module. Continuous evaluation of the results worked	33.33%	0	0	1, 5, 4, 8, 7, 14, 11, 12, 13, 15, 6
Quantitative Genetics Module. Continuous evaluation of the results worked	33.3%	0	0	2, 8, 7, 11, 12, 13, 15, 16, 6

Bibliography

Genomics Module

Included in the practice guideline that is available to students on the Virtu

Human Genetics Module

- Genetic Variation: a laboratory manual. Edited by M.P. Weiner, S. B. Gabriel, J C. Stephens (2007). Cold spring Harbor Laboratory Press, New York, USA.
- Molecular Cytogenetics. Protocols and Applications. Edited by Y-S. Fan (2002). Humana Press Inc., Totowa, New Jersey, USA.

- Human Cytogenetics. Constitutional Analysis. Edited by D.E. Rooney (2001). Third edition. Oxford University Press, Oxford, UK.
- Human Cytogenetics. Malignancy and Acquired Abnormalities. Edited by D.E. Rooney (2001). Third Edition. Oxford University Press, Oxford, UK.

Quantitative Genetics Module

Included in the practice guideline that is available to students on the Virtual Campus.

Software

MEGA (https://www.megasoftware.net/)

PQGen (P6)	https://sites.google.com/a/unizar.es/pqgen/
PLINK (P2 y P3)	http://pngu.mgh.harvard.edu/~purcell/plink/
R (P4 y P5)	
Genup (P6)	http://www-personal.une.edu.au/~bkinghor/genup.htm
Dcbsp (P1)	The author of the software (Joaquim Casellas) will provide it before the class.