

**Laboratory IV**

Code: 101944  
ECTS Credits: 3

| Degree           | Type | Year | Semester |
|------------------|------|------|----------|
| 2500890 Genetics | OB   | 2    | 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: Yes  
Some groups entirely in Spanish: Yes

**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 IV is the fourth 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 IV has as objectives the acquisition of experimental skills in 4 specific modules of content:

- Population Genetics
- Mutagenesis
- Databases and basic programming
- Developmental Biology
- Scientific documentation

**OBJECTIVES**

**Module of Population genetics**

Two are the main objectives of this module: (1) understanding the factors that modulate the genetic variation in the populations and (2) learn how to describe the nucleotide variation of a gene. For the first objective the POPULUS 5.4 software package will be used, which allows to simulate different population processes and

provides a graphical representation of the results. For the second, students will employ the Polymorphism Diversity Analysis Software (PDA) developed by researchers at the Genetics Unit.

#### Module of Mutagenesis

The main objectives of the Mutagenesis module are: To carry out a small biomonitoring pilot study in humans by determining the induction and origin of the micronuclei observed in desquamation cells of the oral mucosa, and to find out their possible relationship with polymorphisms of the glutathione-S transferase gene. In addition to learn how to follow the protocols and use the required experimental methodologies for the study, students will become familiar with the statistical analysis of the results obtained using the PSPP software.

#### Module of Databases and basic programming

The current scenario of research in the genetic field requires that the researcher know and use computer tools. How genetic information is stored in databases? How can you extract this information flexibly? How to create programs to manipulate and analyze genetic data?

The objectives of the module are to create programs and use database management systems that are currently being used in the genetic field.

- Learning to program with the Python language
- Managing and consulting databases
- Applying this instrumental capacity to the processing of genetic data
- Promoting the connection between the computer tools and the genetic data, a key aspect of the bioinformatics research

#### Module of Scientific Documentation

This module aims to provide a general theoretical and practical basis to deep into the use of information resources specialized in genetics and related disciplines. The specific objectives of the module are:

Learn how to solve information needs in the field of genetics and related sciences through the use of bibliographic resources.

Know how to propose strategies for the search and retrieval of information on electronic sources.

### Competences

- Adapt to new situations.
- Apply scientific method to problem solving.
- Define mutation and its types, and determine the levels of genic, chromosomal and genomic damage in the hereditary material of any species, both spontaneous and induced, and evaluate the consequences.
- 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.
- 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.
- Take the initiative and demonstrate an entrepreneurial spirit.
- 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.
- Use and manage bibliographic information or computer or Internet resources in the field of study, in ones own languages and in English.
- Work individually and in teams.

### Learning Outcomes

1. Adapt to new situations.

2. Analyse and interpret human genetic variation.
3. Apply available software programs and web applications to the editing, comparison and structural, functional and evolutionary interpretation of sequences of nucleic acids and proteins.
4. Apply different methods of phylogenetic analysis to molecular sequences to make phylogenetic reconstructions and epidemiological interpretations.
5. Apply scientific method to problem solving.
6. Apply the basic common techniques of a human genetics laboratory.
7. Describe and interpret genetic diversity in different functional regions of the genome.
8. Design and apply in vitro DNA amplification technologies by means of a polymerase chain reaction (PCR) in different specific contexts.
9. Detect the nature of genic mutations and determine the phenotypical consequences.
10. Determine the nature of chromosomal disorders and their effects by employing in situ hybridisation and chromosome painting techniques.
11. Evaluate the mutagenicity of chemical compounds.
12. Extract and purify DNA in human and animal samples.
13. Perform in vivo and in vitro mutagenesis.
14. Simulate and interpret the individual and overall effect of different genetic factors in populations on genetic polymorphism and molecular evolution.
15. Simulate processes of stochastic extinction applied to the genetic conservation of endangered species.
16. Take the initiative and demonstrate an entrepreneurial spirit.
17. Use and manage bibliographic information or computer or Internet resources in the field of study, in ones own languages and in English.
18. Work individually and in teams.

## Content

### Module of Population Genetics

The Population genetics module is organized in 5 sessions of 2.5 hours each and they will be held in the computer room.

Practice 1-3. Simulations by computer with the program POPULUS. Study of the interaction of the main forces that modulate the evolution.

|         |    |           |     |           |
|---------|----|-----------|-----|-----------|
| Session | 1. | Selection | and | drift.    |
| Session | 2. | Selection | and | mutation. |

Session 3. Population structure: selection and migration.

Practice 4. Molecular genetics of populations. Analysis of the nucleotide in the G6pd gene diversity

### Module of databases and basic programming in Pearl

The Module of Databases and Programming Fundamentals consists of 4 sessions of 2/3 hours each. The classes will be held in the computer room, where the theoretical contents of the Module Instrumental Techniques will be applied.

- Session 1: Databases management with MySQL database manager (3h)
- Session 2: Programming in Python. Basic operations (3h)
- Session 3: Programming in Python. Flow control and files (2h)
- Session 4: Programming in Python. Functions and regular expressions (2h)

### Module of Mutagenesis

Scheduled practices in these sessions will allow the student to learn the basic techniques of:

- Practice 1: preparation of miconuclei in buccal mucosa cells
- Practice 2: DNA extraction

- Practice 3: preparation and staining observation of micronuclei
- Practice 4: Electrophoresis gels and REAL-TIME PCR
- Practice 5: Completion of questionnaires, annotation of the data and statistical analysis of the results.

#### Module of Developmental Biology

The module of Developmental Biology is organized in two sessions: a first session of 2.5h at the laboratory, and a second session of 1.5h at the computer lab.

Session 1: Handling, observation of embryos and expression patterns of *Drosophila*.

Session 2: Analysis of expression patterns of *Drosophila* from databases.

#### Module of Scientific Documentation

The module of scientific documentation is divided in 2 sessions of 2 hours each, to be held in computerized classrooms. The contents of each session will be:

1. Bibliographic resources specialized in technical and scientific information: publishing Portals (SCOPUS, Science Direct); Web of Science. Databases in the field of medical distributor (ProQuest).
2. Bibliographic resources provided by the National Library of Medicine: Medline, Pubmed, GenBank, OMIM. Access to information about patents: Latipat and Espacenet.

### 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.

#### Genetic Module of Populations

Autonomous learning through which the student learns to work and is guided at all times by the responsible professor that establishes the objectives of the practice. Students must elaborate, interpret the results obtained and respond to the different questions raised in the practice manual.

#### Module Mutagenesis

The students will have a detailed script 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 script 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 as an example of an exposed group.

#### Database Module and Programming Funds

Students will have an interactive practice guide (Jupyter Notebook) that they will consult to carry out the practices. To facilitate the understanding of the contents and a good development of classes it is recommended that the student read the practice guide before each session. It is also recommended to have the notes of the corresponding theory subject available.

During the session, questions and issues will be solved and at the end of each session the Jupyter Notebook will be proposed to deliver. In the end of the third practical class an integrative program must also be delivered which intends to work and integrate on all the aspects seen during the practices.

## Module Developmental Biology

Students will have a Laboratory Hand-out with the protocols and information to perform the practicals with support from the professor. The hand-out will be available at the Campus Virtual of the course.

At the laboratory, students will learn the basic techniques for handling and visualizing embryos, using *Drosophila* as a model. At the computer lab students will work with specific databases and use bioinformatics tools for their analysis.

## Module Scientific Documentation

Each session will include a first part of theoretical exposition and a second of a practical type where the student will have to perform exercises related to the query of sources and the resolution of informational needs. The questions to be resolved will always be presented contextualized within the thematic field of genetics and related sciences.

Before each session, in the Campus Virtual, materials related to the contents that will be discussed in class will be published. It is advisable for the student to do a reading and a previous review in order to get the most out of the session.

## Activities

| Title                                                    | Hours | ECTS | Learning Outcomes              |
|----------------------------------------------------------|-------|------|--------------------------------|
| Type: Directed                                           |       |      |                                |
| 56/5000 Module Databases and Fundamentals of Programming | 8     | 0.32 | 1, 2, 4, 7, 16, 14, 18         |
| Module Developmental Biology                             | 6     | 0.24 | 5, 3, 16, 18, 17               |
| Module Mutagenesis                                       | 17.5  | 0.7  | 5, 6, 11, 9, 10, 8, 12, 13, 16 |
| Module Scientific Documentation                          | 4     | 0.16 | 1, 5, 16, 18, 17               |
| Module of Population Genetics                            | 12.5  | 0.5  | 2, 4, 5, 7, 16, 14, 15, 17     |
| Type: Supervised                                         |       |      |                                |
| Individual Tutorials                                     | 1     | 0.04 |                                |
| Type: Autonomous                                         |       |      |                                |
| Laboratory booknote                                      | 2     | 0.08 |                                |
| Study                                                    | 16    | 0.64 |                                |

## 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.

#### Genetic Module of Populations

This module will be evaluated by answering questions from a questionnaire after each practical session (50%) and a final exam (50%).

#### Module Mutagenesis

The module's mark will be obtained averaging an exam (70%) and a questionnaire related to practical sessions (30%). In order to make the average between the two evaluations and complete the module, each mark must be graded  $\geq 5$  independently.

#### Module Databases and Fundamentals of Programming

The module's grade will be obtained through a final exam (50%) and the exercises carried out in each session that must be delivered to the teacher together with the student's attitude in the classroom (50%). To be able to make the average between the two parts, it will be necessary to have a note  $\geq 4$  in each part independently.

If you do not reach the minimum mark to be able to do the average, each of the parts will be recovered independently. The part of the final test will be recovered on the scheduled date for the recovery of the subject. In the case of not having passed the exercises, the teacher will inform the procedure and deadlines.

#### Module Developmental Biology

This evaluation of this module will be through: (1) a short report integrating the results obtained in both sessions (80%), and (2) the student attitude and work during the practical sessions, both at the wet lab and the computer lab (20%)

#### Module Scientific Documentation

Assistance to sessions (weight = 20%): attendance is mandatory. Each unjustified absence will entail a penalty of 0.5 points, within this concept.

Proof of knowledge about the theoretical and practical contents (weight = 80%): it will include short answer questions as well as information search problems that the student will have to solve through the use of the computer.

To pass the course, it is necessary to first approve each module with a mark  $\geq 5$ .

Students who do not pass the different modules of the subject or wish to improve their mark can present themselves on the scheduled date for the recovery evaluation of the subject. The presentation of the student to the examination of improvement of note implies the renunciation of the qualification obtained previously. 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 the specific 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.

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 module.

### Assessment Activities

| Title | Weighting | Hours | ECTS | Learning Outcomes |
|-------|-----------|-------|------|-------------------|
|-------|-----------|-------|------|-------------------|

|                                                                                                             |       |     |      |                               |
|-------------------------------------------------------------------------------------------------------------|-------|-----|------|-------------------------------|
| Module Databases and Fundamentals Programming Questionnaires<br>Continuous evaluation of the results worked | 22,5% | 1.8 | 0.07 | 1, 3, 16, 18                  |
| Module Developmental Biology Continuous evaluation of the results worked                                    | 22,5% | 1.8 | 0.07 | 4, 5, 3, 16, 15, 18, 17       |
| Module Mutagenesis Questionnaires Continuous evaluation of experimental results                             | 22,5% | 1.8 | 0.07 | 5, 11, 9, 10, 13, 16          |
| Module Scientific Documentation                                                                             | 10%   | 0.8 | 0.03 | 1, 5, 16, 18, 17              |
| Module of Population Genetics. Questionnaires Continued assessment of the results worked                    | 22,5% | 1.8 | 0.07 | 2, 5, 6, 7, 8, 12, 16, 14, 17 |

## Bibliography

### Genetic Module of Populations

Included in the practice manual that is available to students on the virtual campus

### Module Mutagenesis

The practice guide can be downloaded from the Virtual Campus

### Module Databases and Fundamentals of Programming

The practice guide can be downloaded from the Virtual Campus

### Module Developmental Biology

The practice guide can be downloaded from the Virtual Campus

### Module Scientific Documentation

ABADAL, E.; CODINA, LI. Bases de datos documentales: características, funciones y método. Madrid: Síntesis, 2005.

ADLER, David A. "Human genetics: online resources" [En línea]. Encyclopedia of life sciences, 2001. [Consulta: 12-07-2014]. Disponible a: [bit.ly/1oXnTV6](http://bit.ly/1oXnTV6)

ALEIXANDRE, R. "Fuentes de información en ciencias de la salud en Internet" [En línea]. Panacea@, 2011, vol. 11, núm. 33. [Consulta: 11-07-2014]. Disponible a:

<http://www.medtrad.org/panacea/IndiceGeneral/n33-Ponencias-Aleiandre.pdf>

CORDÓN, J.A, et al. Nuevas fuentes de información: información y búsqueda documental en el contexto de la web 2.0. Madrid: Pirámide, 2010.

Fuentes de información biomédica [En línea]. Cedimcat. [Consulta: 09-07-2014]. Disponible a:

<http://www.cedimcat.info/html/es/dir2471/doc26734.html>

NATIONAL HUMAN GENOME RESEARCH INSTITUTE. Online Genetics Education Resources [En línea].

National Institutes of Health. [Consulta: 11-07-2014]. Disponible a: <http://www.genome.gov/10000464>