

2019/2020

### **Genetics**

Code: 100891 ECTS Credits: 6

Degree	Туре	Year	Semester
2500252 Biochemistry	FB	2	1

#### Contact

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# **Use of Languages**

Principal working language: spanish (spa)

Some groups entirely in English: No
Some groups entirely in Catalan: No
Some groups entirely in Spanish: No

## Other comments on languages

Even though the linguae francae of this course are Spanish and Catalan, part of the study material is in English

### **Prerequisites**

There are no official prerequisites. Even so, a basic knowledge of probability, combinatorics, and statistics is recommended. On the other hand, to ensure the student will properly follow the classes and achieve the learning outcomes proposed, it is essential to know the cellular processes studied in cytology, with particular emphasis on the cell cycle, mitosis, and meiosis.

#### **Objectives and Contextualisation**

The overall objective of this course is that students receive a general introduction to the basic principles of Genetics to understand the laws of heredity, its cytological and molecular basis, and its variation at the population level.

The training objectives are as follows:

- 1) Understand the need for the study of genetics in the context of biochemistry.
- 2) Know the laws of the transmission of genetic information, the chromosomal theory of inheritance, and how to make genetic maps and interpret pedigrees.
- 3) Know the structure, organization and function of the genetic material.
- 4) Know how to use and interpret genomic data.
- 5) Know the main sources of genetic variability in populations.

#### Competences

- Be able to self-evaluate.
- Collaborate with other work colleagues.

- Display knowledge of the biochemical and genetic changes that occur in many pathologies and explain the molecular mechanisms involved in these changes.
- Interpret experimental results and identify consistent and inconsistent elements.
- Manage bibliographies and interpret the information in the main biological databases, and also know how to use basic ICT tools.
- Manage information and the organisation and planning of work.
- Stay abreast of new knowledge of the structure, organisation, expression, regulation and evolution of genes in living beings.
- Take responsibility for one's own learning after receiving general instructions.
- Think in an integrated manner and approach problems from different perspectives.
- Understand the language and proposals of other specialists.
- Use ICT for communication, information searching, data processing and calculations.

### **Learning Outcomes**

- 1. Analyse a pedigree thoroughly and define the types of inheritance of a particular genotype and phenotype.
- 2. Be able to self-evaluate.
- 3. Calculate data related to physiological processes in animals.
- 4. Collaborate with other work colleagues.
- 5. Describe genetic alterations that can be found to underlie certain pathologies.
- 6. Describe the determining factors in evolution.
- 7. Explain the fundamental principles of genetics and reproduction.
- 8. Interpret experimental results and identify consistent and inconsistent elements.
- 9. Manage information and the organisation and planning of work.
- 10. Solve practical problems in genetics (including population genetics).
- 11. Take responsibility for one's own learning after receiving general instructions.
- 12. Think in an integrated manner and approach problems from different perspectives.
- 13. Understand the language and proposals of other specialists.
- 14. Use ICT for communication, information searching, data processing and calculations.

### Content

- Topic 1: Genetics. Fundamental concepts. Genetic analysis. Model organisms.
- Topic 2: Mendel's principles of inheritance.
- Topic 3: Genetic consequences of mitosis and meiosis. Chromosomal theory of inheritance. Biological cycles and reproduction.
- Topic 4: Sex-linked inheritance patterns. Sex determination. Family trees analysis and genetic counseling. Dosage-compensation mechanisms.
- Topic 5: Extensions of the Mendelian inheritance. Multiple allelism. Lethal genes. Genotypic interactions. Epistasis. Biochemical genetics. Penetrance and expressivity.
- Topic 6: Non-Mendelian Inheritance. Cytoplasmic inheritance: mitochondria and plastids. Mobile genetic elements.
- Topic 7: Genetic linkage and recombination in eukaryotic organisms.
- Topic 8: Genetic maps. Crossbreeding. Cytological demonstration of cross-over. Mitotic recombination. Genetic maps in humans.
- Topic 9: The double helix and the flow of genetic information. Genes in action. Fundamentals of replication, transcription and translation. The genetic code.

Topic 10: Mutations. Spontaneous mutation and induced mutations. Types of mutations. Main mutagenic agents. Repair mechanisms.

Topic 11: Numerical and structural chromosome changes. Chromatid and chromosome breaks. Deletions. Duplications. Inversions. Translocations. Variations in the number of chromosomes: euploidy and aneuploid. Aneuploidy in humans. Polyploidy: Auto- and allopoliploids.

Topic 12: Quantitative inheritance. Multilocus traits. Meaning of polygenic inheritance. Heritability. Variation partitioning. Measures of heritability. Quantitative inheritance in humans. Studies in twins.

Topic 13: Population genetics. Mendelian populations. Gene and genotypic frequencies. Hardy-Weinberg's law. Selective mating. Forces of evolution: mutation, genetic drift, migration and natural selection. The selection of quantitative characters.

### Methodology

The teaching methodology is based on two approaches: a theoretical and a practical one.

#### Theoretical approach

Classes will give the students the basic knowledge needed to understand the course's contents. Support material will be available on the virtual campus. It is recommended that students take the presentations published in the CV to classes, in order to follow them easily and take notes, if necessary. The concepts explained in class will have to be autonomously deepened to promote the development of non-guided learning strategies. In order to facilitate this task, bibliography, audiovisual and interactive material will be provided. Finally, individual tutorials are planned for students who wish to do so. These tutorials should be used to potentiate the students' progress and to help them understand the most difficult or complex concepts.

### Practical approach

Problem seminars will be used to learn how to apply the previously-acquired knowledge. Students will find the problems that will be treated on each seminar on the Virtual Campus. Seminars will be given in reduced groups and will be based on the discussion and the resolution of practical problems, applying theoretical concepts together with mathematical tools and statistics. Students who want it can request individual tutorials to facilitate the understanding of the most complex problems.

#### **Activities**

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Group tutorials	3	0.12	8, 12
Lectures	30	1.2	1, 5, 6, 7, 9, 8, 12, 11
Problem classes and seminars	10	0.4	14, 3, 8, 10
Type: Supervised			
Individual tutorials	9	0.36	8, 12
Type: Autonomous			
Bibliographic search	9	0.36	14, 8, 11
Consultation of recommended books	8	0.32	9, 8, 12, 11

Problem resolution	15	0.6	1, 3, 4, 8, 12, 10
Study	62	2.48	9, 12, 10, 11, 2

#### **Assessment**

#### **Fvaluation**

The subject will be evaluated by 2 written tests that correspond to both theoretical and practical problems. The quality of the students' work will also be taken into account. The evaluation system considering the specific weight of each part will be as follows:

- 1. Exams. There will be 2 midterm exams to evaluate the progressive understanding and acquisition of the contents. The exams weight 60% of the final mark. To pass the exam it is necessary to have a 5.
- 2. Evaluation of 2 assignments in terms of its presentation, structure, clarity, content and synthesis capacity. This concept represents 40% of the final mark.
- 3. Retake examination. It will correspond to the part or parts not previously passed. It may also serve to improve the mark. A minimum of 5 is needed in every evaluation to pass the course.

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

#### **Assessment Activities**

Title	Weighting	Hours	ECTS	Learning Outcomes
Groupal assignments	40%	0	0	14, 3, 4, 13, 9, 12, 10, 11, 2
Two midterm exams (First: 30%; second: 30%)	60%	4	0.16	1, 3, 5, 6, 7, 9, 8, 12, 10

### **Bibliography**

### Theory:

- 1) Pierce, B.A. 2016. Genetics. A conceptual approach. (5th edition). Ed. Médica Panamericana.
- 2) Pierce, B.A. 2011. Genetic Essentials. Concepts and Connections. Ed. Médica Panamaericana.
- 3) Benito, C. & Espino, F.J. 2013. Genética. Conceptos esenciales. Ed. Médica Panamericana.

#### Problems:

- 1) Elrod, S.L. & Stansfield, W.D. 2010. Schaum's Outline of Genetics. Fifth edition. Mc Graw-Hill, USA.
- 2 Jiménez, A. 2008. Problemas de Genética para un curso general. (3rd edition). Colección Manuales UEX. University of Extremadura.
- 3) Ménsua, J. L. 2003. Genética. Problemas y Ejercicios resueltos. Pearson Prentice Hall, Madrid.

It is recommended to consult the teaching space of the subject.