

**Genetics**

Code: 101963  
ECTS Credits: 6

Degree	Type	Year	Semester
2500890 Genetics	OB	1	2

## Contact

Name: Antoni Barbadilla Prados

Email: antonio.barbadilla@uab.cat

## Teaching groups languages

You can check it through this [link](#). To consult the language you will need to enter the CODE of the subject. Please note that this information is provisional until 30 November 2023.

## Teachers

Javier Gutierrez Garcia

Cristian Valiente Gil

## Prerequisites

- The own requirements of access to the degree.
- It is convenient that the student review the basic processes of cell division of the subject of Cellular Biology and Histology
- The foundations of probability that have been seen in the subject of Mathematics, and know the statistical distributions binomial, Poisson and normal.
- In order to attend the practical sessions it is necessary that the student justifies having passed the biosafety and safety tests that will find in the Virtual Campus and to be knowledgeable and accept the working rules of the laboratories of the Faculty of Biosciences.

## Objectives and Contextualisation

The subject of Genetics is taught in the 1st year of the Degree of Genetics (2nd semester). It is the first subject with specific content of genetics, and that is why they will provide the basic foundations of the hereditary transmission, that is, how genetic information is transferred between generations in both individuals and populations. Molecular genetics is looked very briefly, since it will be treated in depth in the second course. The contents of this course include the transmission of chromosomes and genes, the development of genetic maps, the mutation of the genetic material, the inheritance of characters of continuous variation, the genetics of populations, and the genetic properties and characteristics of the model organisms.

Each class of this course wants to be a unique opportunity to meet the new and fascinating ideas of Genetics. From the very beginning we want to convey an overview of the problems and the scope of this Science. We

want to motivate you, creating a contagious interest and enthusiasm, promoting critical inquiry and developing curiosity about the issues of genetics. We are fortunate that we can talk about the excitement and vitality of this science realistically, without the need to pretend. The subject wants to be a lasting guide, a continuous reference, to which you can turn mentally over and over again.

From the point of view of learning to acquire, students are expected to understand the power of inquiring of the genetic analysis, which has opened the door to many of the great discoveries of genetics, and how this powerful methodological tool is applied in current genetics. It is also intended to acquire a historical perspective of the great milestones of genetics, from the experiments of Mendel to the sequencing of the human genome.

As a complement to face-to-face training this course has an online learning platform that implements the new and powerful learning and knowledge technologies (LKT) in the course. This resource aims to facilitate personalized work, individual discovery, integration of different sources of information, as well as enhancing originality and the development of innovative skills, all with the ultimate goal that the student acquires new perspectives for the construction and understanding of the knowledge and skills required for the training of a professional geneticist. Many of the exercises and tasks to be done during the course emphasize the multidisciplinary nature of Genetics. Among other tools, the online Portfolio of works, tasks and activities either assigned or self-created by the student, is a featured element for the monitoring and evaluation of the learning acquired by the student throughout the course.

The formative objective is to acquire a solid understanding of the bases and mechanisms of inheritance and the method of genetic analysis: to be able to explain and interpret the principles of the transmission of genetic information, to analyze genealogies and to apply it to genetic counseling, to elaborate and work with genetic maps, to understand what is and how genetic variation is measured in populations, to design and obtain relevant information from genetic experiments and to interpret the results obtained. Explanations will be contextualized historically for the student to visualize how the conceptual building of genetics has been constructed and to appreciate the importance of the current moment of this science, vibrant and full of promises and challenges.

## Competences

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
- Apply scientific method to problem solving.
- Be able to analyse and synthesise.
- Be able to communicate effectively, orally and in writing.
- 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.
- Describe and interpret the principles of the transmission of genetic information across generations.
- Enunciate and evaluate the biological properties and genetic characteristics of model genetic organisms.
- 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.
- Perceive the strategic, industrial and economic importance of genetics and genomics to life sciences, health and society.
- Produce and work with genetic maps.
- 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 scientific method to problem solving.
3. Be able to analyse and synthesise.
4. Be able to communicate effectively, orally and in writing.
5. Describe and interpret the rules for transmitting ligated genes.
6. Describe the different concepts that relate genotype with phenotype.
7. Determine the genetic basis of a character from inheritance patterns.
8. Elaborate genetic maps from crossings of two and three points.
9. Enunciate and evaluate the biological properties and genetic characteristics of model genetic organisms.
10. Estimate the genetic parameters of a character from crossbreeding.
11. Explain and interpret Mendel's experiments and the derived laws of inheritance.
12. Explain that in the past genetics was used unlawfully to foster racist ideologies.
13. Explain the nature of genetic variation, its origin and maintenance in panmictic populations.
14. Identify chromosomal variants and anomalies.
15. Reason critically.
16. Synthesise, based on historical progress in genetics, a perspective of the current and future scope of this science.
17. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
18. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
19. Use and manage bibliographic information or computer or Internet resources in the field of study, in one's own languages and in English.

## Content

Theoretical contents\*

### Part I. Introduction

Topic 1: The science of genetics. Fundamental concepts. Genetic analysis. Model organisms of genetics.

### Part II. Mendelism

Topic 2: Mendelian Principles. Equal segregation and independent assortment. Types of inheritance. Examples of Mendelian inheritance in humans.

Topic 3: Mitosis and meiosis. Chromosomal theory of heredity. Biological cycles.

Topic 4: Inheritance of sex. Determination of sex. Inheritance linked to sex. Inheritance influenced by sex. Inheritance limited to one sex. Compensation of dosis.

Topic 5: Analysis of genealogies and genetic counseling.

Topic 6: Extensions of the Mendelian analysis. Relations of dominance. Multiple Allelism. Lethal allele and essential gene. Penetrance and expressiveness. Genotypic interactions. Epistasis. Biochemical genetics. Hypothesis a gene-an enzyme. Test of complementation.

### Part III. Recombination and genetic maps

Topic 7: Linkage, crossing-over and recombination.

Topic 8: Genetic maps. Genetic mapping: two points cross; three points cross. Cytological and nucleotide demonstration of crossing-over. Analysis of tetrads. Mitotic recombination. Genetic maps in humans.

## Part IV. Quantitative inheritance and non-Mendelian inheritance

Topic 9: Quantitative inheritance. Traits controlled by several loci. Meaning of polygenic inheritance. Heritability. Metrics of heritability.

Topic 10: Non-Mendelian inheritance. Cytoplasmic inheritance: mitochondria, chloroplasts. Transposable genetic elements.

## Part V. DNA and mutation

Topic 11: The double helix.

Topic 12: Mutation. Spontaneous mutation and induced mutation. Types of mutation. Repair.

Topic 13: Numerical and structural chromosomal changes. Deletions and Duplications. Inversions and their effects. Translocations. Variation in chromosome number: euploidy and aneuploidy. Aneuploidy in man. Polyploidy: self and allopolyploidy.

## Part VI. Population genetics

Topic 14: Population genetics. The Mendelian population. Allelic and genotypic frequencies. Hardy-Weinberg equilibrium. Non-random mating. Evolutionary factors: mutation, migration, genetic drift and natural selection

## Contents of the lab practices

Session 1. Introduction to the biology and morphology of *Drosophila melanogaster* (1 session) (Integrated Laboratories)

Session 2. Analysis of a mutant and assignment to its linkage group (1 session) (Integrated Laboratories)

Session 3. Elaboration of a genetic map of three markers (1 session) (Integrated Laboratories)

Session 4. Observation of chromosomes and mutations (somatic recombination, chromosomal alterations, micronuclei) (1 session) (Integrated Laboratories)

## Methodology

The teaching methodology includes six types of activities: theory magistral classes, problem solving, seminar sessions, practical classes, carrying out activities and tasks through the online learning platform and tutoring sessions. This course has an online learning platform ([http://genetica.uab.cat/genetica\\_genetica](http://genetica.uab.cat/genetica_genetica)) to prompt the communicative and collaborative power offered by the technologies associated with the cloud and Web 2.0 to get new ways of working, thinking and learning the science of genetics.

Theoretical classes: Classes to transmit the basic concepts and information necessary to develop autonomous learning. Promotion of the active participation of students through the approach of reciprocal questions. Support of the presential lessons with multimedia material (PowerPoint presentations, animations, quiz, web pages and online videos) that will be available to the student on the [Web de recursos del curso](#).

Problems and applications of self-learning: Resolution and discussion of previously worked problems of autonomous form by the students (the problems are in the page of exercises of the Web of the course ([página de ejercicios](#)) and in the Moodle application Permanent classroom of Genetics) . Students must practice with the self-learning web tool Genetic Permanent Classroom, which has been created by professors from the genetics unit. Through a series of tests, the application allows evaluating the knowledge acquired by the student and assesses both their level of understanding and their shortcomings. Finally, the aspects that the student should work on are advised. Active participation in solving problems and using this tool counts 10% of the final grade. Small groups of 30 students.

Seminars: Groups of 30 or less students. There will be a program of activities ([programa de actividades](#)) that will be carried out individually and/or in groups that can be presented orally and/or as a writing. The teacher

proposes a discussion topic or task to be carried out that the students must solve with the resources that are put at their disposal. Current issues and classic works of genetics will be discussed (Mendel's work and the discovery of the double helix among others). The works must always be accompanied by the references consulted. In the case of group work, the contribution of each one must be specified (for example, "we have all contributed equally"). You can not present the work of another as your own work in any way. Any piece of information that has not been prepared by the student, that is, that has been copied literally from external sources or from other colleagues, must be explicitly indicated in the work. Students can participate in the evaluation of their classmates' work (peer evaluation). The works can be compulsory or optional and should preferably be uploaded in pdf format in the corresponding portfolio of the course website. Originality, argumentative capacity, and the selection of information sources will be valued.

Practices: Laboratory practices in groups of 20 students. We will work with the species *Drosophila melanogaster* as a model paradigm model of genetics. A genetic map and visualization of phenotypic and chromosomal mutants will be carried out. See the Practices section. The students have a script of practices that will be found in the virtual campus within the material of the subject. You have to read carefully the part corresponding to each session before starting the practice in order to get the most out of it. At the end of each session there will be an evaluation test of the practice ([Prácticas](#)).

Tutorials: Discussion and resolution of doubts/problems by the professor. They will be done individually or in small groups to be agreed between the students and the teacher. It is recommended to do at least one group tutoring before each of the exams for the resolution of doubts.

Online learning platform: The open, collaborative and flexible environment that opens the Web 2.0 technology is an unparalleled resource to energize the student-teacher group inside and outside the classroom. This course has a learning Web platform, a resource that is a complement to face-to-face training and its use is intended that each student adopts an active and personalized role in their learning of genetics and that in turn can contribute to learning collaborative and/or cooperative of the whole class. This learning platform contains administration tools, communication, information, publication, collaboration, activities, exercises, authorship, self-assessment, tutoring and a student portfolio, among others.

(Address online learning platform for genetics [http://genetica.uab.cat/genetica\\_genetica](http://genetica.uab.cat/genetica_genetica))

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			
Laboratory practices	12	0.48	2, 5, 7, 8, 13, 14, 15, 4, 3, 19
Seminars and exercises solving	14	0.56	2, 15, 4, 19
Theory classes	30	1.2	2, 6, 5, 7, 8, 9, 11, 13, 14, 15, 4, 3, 19
Type: Supervised			
Group and individual tutorials	6	0.24	2, 6, 5, 7, 8, 9, 11, 13, 14, 15, 4, 3, 19
Preparation of materials	1	0.04	2, 15, 4, 19
Type: Autonomous			
Bibliography Search	4	0.16	15, 19

Drafting of work and preparation of tasks of the portfolio	14	0.56	2, 15, 4, 19
Readings	8	0.32	15, 19
Resolution of exercises	20	0.8	2, 15, 4, 19
Study	35	1.4	2, 15, 19

## Assessment

The competences of this subject will be evaluated through continuous evaluation, which includes different activities: two exams, a final 2n exam or improvement of grades, written tests, problem solving, laboratory work and participatory activity in the classroom.

The evaluation system is organized into 5 evaluation activities, each assigned a specific weight in the final grade:

Evaluation of laboratory practices: the laboratory notebook and the answers to the questionnaires at the end of each practice will be evaluated. Overall weight of 20%.

Evaluation of the resolution of problems and the use of the "permanent genetics classroom" tool. Overall weight of 10%.

Evaluation of the tasks, presentations and participation in the seminars: this activity has a global weight of 15%.

Written exams: Two partial exams and a final recovery exam (or to improve the grade). The tests are combined, and consist of questions written answers, problem solving and test type. This section will have a global weight of 60%. The first partial will have a weight of 25% and the second of 30% of the overall score. The weight of the second written evaluation is greater than that of the first because it also includes the contents of the first evaluation. To pass the subject you must reach a minimum grade of 4.0 in each of the exams and the average of the marks of both partials must be  $\geq 5.0$ . (Exam models)

Evaluation of the online portfolio: tasks and optional activities or student's own creation work. The student portfolio shows in a tangible way the performance and progress, the achievements and the understanding that the student has or has reached along the course of the matter. This evaluation can add up to 1.0 points to the final grade.

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 weight of all conducted evaluation activities is less than 67% of the final score.

Final grade weighting formula:

Final grade = [Exams (partial or final)] x 0.55 + (Problems and Permanent Classroom) x 0.10 + (Seminar activity) x 0.15 + (Practice assessment) x 0.20 + Optional work uploaded to portfolio (up to 1.0 points maximum).

The subject is considered passed if the final grade is  $\geq 5.0$ .

The maximum final grade that can be achieved is 10.

Students who, having passed the partial theory tests and/or problems, want to improve their grade may choose to take the final test for the entire subject or for one of the partial tests. This test will be different from the recovery test. The note of the final test will be the one that will prevail.

Not Assessable

Students will obtain the grade "Not Assessable" when the evaluation activities carried out have a weighting of less than 67% in the final grade.

## Unique evaluation

The students who take advantage of the unique evaluation will take a single synthesis test in which the contents of the entire theory program of the subject will be evaluated. The test will consist of theoretical questions and problems and will take place coinciding with the same date set in the calendar for the last continuous assessment exam.

The same evaluation system will be applied as for the continuous evaluation. The grade obtained in this synthesis test will account for 55% of the final grade for the subject.

Laboratory practices (PLAB), seminars and problems (PAUL) are evaluated in the same way as in the continuous assessment. The grade obtained will mean 45% of the final grade for the course (20% laboratory practices, 15% activities and work from seminars, and 10% solving problems and genetic tests).

The students who take advantage of the single evaluation must carry out the laboratory practices (PLAB) in face-to-face sessions and it is a requirement to have them approved with a weight of 20% of the final grade for the subject.

## Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Evaluation of the online portfolio of optional tasks or self-created works	Until 10% plus	0	0	1, 18, 17, 2, 15, 4, 3, 19
Final exam (individual grading)	Two part exams (individual grading)	2	0.08	1, 18, 17, 2, 6, 5, 7, 8, 9, 10, 11, 13, 14, 15, 4, 3, 19
Final exam (individual grading)	55% (25% and 30%)	4	0.16	1, 18, 17, 2, 6, 5, 7, 8, 9, 10, 11, 13, 12, 14, 15, 4, 16, 3, 19
Participation and realization of the activities programmed in the seminars	15%	0	0	1, 18, 17, 2, 12, 15, 4, 16, 3, 19
Problem solving and use of the tool Aula permanente de genètica	10%	0	0	2, 6, 5, 7, 8, 11, 13, 15, 4, 3, 19
Questionnaires for laboratory practices (individual evaluation)	20%	0	0	2, 6, 5, 7, 8, 9, 11, 13, 14, 15, 4, 3, 19

## Bibliography

Theory:

- Pierce, B. A. (2016). Genética: [Un enfoque conceptual. 5a Edición. Editorial Panamericana.](#)
- Benito, C & Fco. Javier Espino (2013). [Genética. Conceptos esenciales. Editorial Panamericana.](#)
- Griffiths, A.J.F., S.R. Wessler, R.C. Lewontin & S.B. Carroll (2008). [Genética \(9a edición\). McGraw-Hill/Interamericana.](#)
- Moltó, M.D. & L. Pascual. (1999). [Però, què és això de la genètica? Ed. Universitat de València.](#)

- Pierce, B. A. (2011). [Fundamentos de Genética: Conceptos y relaciones](#). Editorial Panamericana.
- [More references on Fundamentals of Genetics](#)

Problems:

- Benito C. 2015. 141 problemas de genética: Resueltos paso a paso. Editorial Síntesis. Madrid.
- Elrod, S. & Stansfield, W.D. 2002. Schaum's Outline of Genetics. Fourth edition. Mc Graw-Hill, USA.
- Ménsua, J.L. 2003. Genética. Problemas y ejercicios resueltos. Pearson Prentice Hall, Madrid.

Assay on Genetics:

- Siddhartha, M. 2017. [El Gen: a personal account](#) - Editorial debate.

Course Web page:

- Plataforma Web 2.0 para la docencia del curso <http://genetica.uab.cat>
- Aula permanente de genética <http://genetica.uab.cat/aulagenetica>

## Software

Course Web site:

- Web 2.0 Platform for genetics course teaching <http://genetica.uab.cat>
- Permanent Classroom in Genetics <http://genetica.uab.cat/aulagenetica>
- Genetics - Campus Virtual UAB <https://e-aules.uab.cat/2021-22/course/view.php?id=41822>