

**COURSE DATA****Data Subject**

<b>Code</b>	33047
<b>Name</b>	Genetics
<b>Cycle</b>	Grade
<b>ECTS Credits</b>	10.0
<b>Academic year</b>	2022 - 2023

**Study (s)**

<b>Degree</b>	<b>Center</b>	<b>Acad. Period year</b>
1100 - Degree in Biology	Faculty of Biological Sciences	2 Annual

**Subject-matter**

<b>Degree</b>	<b>Subject-matter</b>	<b>Character</b>
1100 - Degree in Biology	7 - Molecular and genetic basis of living beings	Obligatory

**Coordination**

<b>Name</b>	<b>Department</b>
PASCUAL CALAFORRA, LUIS FCO.	194 - Genetics
SILVA MORENO, FRANCISCO J.	194 - Genetics

**SUMMARY**

The course of Genetics is taught in second course curriculum Degree in Biology from the University of Valencia. It's part of the *Molecular and genetic material of living beings*, which is composed of three subjects. Two are 10 ECTS each, *Biochemistry* and *Genetics*, while the third, *Molecular Biology Methods* is 6 ECTS. The course of *Genetics* is theoretical and practical and will be held throughout both semesters that make up the academic year.

The delivery of the contents of molecular genetics / molecular biology has been coordinated with the other two subjects with special attention to developing a coordinated program of activities and contents in order to avoid overlaps. The objectives related to the acquisition of practical skills will be especially shared and supplemented with the subject of *Molecular Biology Methods* as this course aims to integrate different disciplinary-cellular molecular techniques, several of which have a clear connection to our area knowledge.



In addition, continuing the process of coordination of content between subjects, some aspects of evolutionary genetics are included in the subjects *Tree of Life* (6 ECTS), first course, and *Evolutionary Processes and Mechanisms* (4.5 ECTS) given during the first semester of the second course, while not the same, therefore, between the contents of the subject of *Genetics*.

The general objectives of the course are to provide the student genetics on the one hand, basic knowledge concerning the study of biological variability and the mechanisms that regulate their heritage, and the structure and function of genes and genomes, and other tools conceptual and methodological enable it to carry out any type of genetic analysis in their professional work.

## PREVIOUS KNOWLEDGE

### Relationship to other subjects of the same degree

There are no specified enrollment restrictions with other subjects of the curriculum.

### Other requirements

## OUTCOMES

### 1100 - Degree in Biology

- Skills in analysis and synthesis.
- Capacidad de resolución de problemas.
- Capacidad de razonamiento crítico.
- Capacidad de aprendizaje autónomo.
- Capacidad de comunicación oral y escrita.
- Capacidad de manejar el inglés como vehículo de expresión científica.
- Capacidad de utilizar las nuevas tecnologías de información y comunicación.
- Comprender el método científico.
- Capacidad de trabajar en equipo.
- Saber hacer análisis de datos científicos.
- Capacidad de búsqueda de información y análisis crítico de textos científicos.
- Conocer los mecanismos de la herencia biológica.
- Conocer los mecanismos de replicación, transcripción, traducción y modificación del material genético.



- Conocer las bases biológicas del desarrollo.
- Conocer las metodologías de análisis global estructural y funcional de genomas y procesos celulares.
- Conocer la estructura y función de las biomoléculas.
- Conocer los conceptos básicos y las aplicaciones de la tecnología del DNA recombinante y de la Ingeniería Genética.
- Capacidad para trabajar correctamente en los laboratorios de Bioquímica, Genética y Biología Molecular, incluyendo seguridad, manipulación, eliminación de residuos y registro anotado de actividades.
- Capacidad para utilizar la instrumentación básica en los laboratorios de Bioquímica, Genética, Biología Molecular y Celular.
- Tener una visión integrada de las técnicas y métodos utilizados por la Bioquímica, Genética y Biología Molecular.
- Capacidad para diseñar experimentos y aproximaciones multidisciplinares para la resolución de problemas concretos.
- Capacidad para presentar, discutir y extraer conclusiones de los resultados de los experimentos científicos.

## LEARNING OUTCOMES

1. Resolving issues and problems
2. Practical activities in the laboratory and analytical results.
3. Work experience in the computer lab and interpret the results.
4. Conducting group work and written and oral presentation with audiovisual support.
5. Design experiments to solve specific problems.
6. Perform genetic analysis.
7. Manipulating the genetic material.
8. Analyzing and comparing nucleic acid sequences and proteins.
9. Integrating molecular and genetic aspects of cellular organization and function

## DESCRIPTION OF CONTENTS

### 1. Introduction to Genetics

Definition and objectives of genetics.

Basics: genotype, phenotype and standard reaction.

Phenocopies.

Pleiotropy

Genetic analysis.

Mutation: definition and types.

Relations between alleles.



## 2. Patterns of inheritance.

Cytological basis of inheritance, mitosis and meiosis.  
Genetic consequences of meiosis.  
Gregor Mendel: the reasons for success.  
Monohybrid Crossing: Law of segregation.  
The testcross.  
Dihybrid analysis: Law of the transmission.  
Genetic notation.  
The use of diagrams branched and crosstabs.  
The chi-square test.  
Analysis of polyhybridism.  
Study and calculation of probabilities in genealogies

## 3. Extensions of Mendelian analysis

The ABO blood group system, an example of multiple allelism.  
How to establish dominance between alleles series.  
Lethality.  
Relationships between genes: interaction and epistasis.  
Penetration and expressiveness.  
Influence of the environment on gene expression.

## 4. Gene function and complementation

A. Garrod and inborn errors of metabolism.  
G. Beadle, E. Tatum and the beginning of biochemical genetics.  
The genetic dissection of a biochemical pathway.  
Sickle cell anemia: Each gene encodes a polypeptide.  
Gene complementation.  
The flow of hereditary information.

## 5. Genetics of quantitative traits.

The multiple factors.  
Standard reaction and phenotypic distribution.  
Dismembering the phenotypic variance.  
When is worth doing selection?  
Heritability.  
How to calculate the heritability of a character.



## 6. Population genetics

The genetic pool.  
Frequency of genes and genotypes.  
The Hardy-Weinberg law.  
Extensions of the Hardy-Weinberg law.  
How to tell if a stock is in equilibrium.  
Using the Hardy-Weinberg law to estimate gene frequencies.

## 7. Chromosomes, sex and heredity

Establishment of the chromosome theory of heredity.  
Linked inheritance sex chromosomes.  
Dosage compensation.  
Sex determination systems.  
Role of X and Y chromosomes in *Drosophila* and humans.  
Influence of sex on inheritance and gene expression.  
Influence of environment on gene expression.

## 8. Genetic linkage

The transfer of linked genes.  
Meiotic recombination.  
Detection of linkage.  
Recombination, genetic distance and linkage map.  
Mitotic recombination.  
Intragenic recombination.

## 9. Genetic mapping in eukaryotes

The map of three points.  
How to proceed when we do not know the order of genes.  
Genetic distance and physical distance.  
The phenomenon of interference.  
Double crossovers and map functions.  
Distance from the dihybrid.  
Linkage analysis in pedigrees: lod score.  
Segregation and recombination in haploids: tetrad analysis.



### 10. Transferring, recombination and maps of the hereditary material in bacteria and viruses

The transformation and cotransformation maps.  
The bacterial conjugation.  
Characteristics of the factor F.  
Interrupted mating maps.  
Taking advantage of the phage, transduction.  
Cotransducción maps.  
Recombination in viruses.  
Intragenic recombination.

### 11. The nature of the hereditary material: Nucleic acids and Inheritance

Features to be met by the hereditary material.  
The transforming principle of F. Griffith.  
Establishing the nature of the transforming principle.  
RNA is the hereditary material of some viruses.  
Interactions between DNA and proteins.

### 12. The chromosome: inheritance vehicle

Chromatin: composition and organization.  
From the nucleosome to the metaphasic chromosome.  
Centromere, telomere and nucleolar organizer.  
Morphology and chromosome number.  
Karyotype.  
Staining of chromosomes.  
The hybridization "in situ" as a technique for chromosome identification.  
Euchromatin, heterochromatin and position effect.

### 13. Chromosomal mutations

A classification of chromosomal mutations.  
Structural changes.  
Duplications and deletions affect the number of genes in the chromosomes.  
Inversions and translocations change the physical location of genes.  
Numerical changes.  
Chromosomal fusion and fission: Robertsonian translocations.  
Aneuploidy a partial change in chromosome sets.  
Polyploids: a variation of the euploidia.



#### 14. Physical maps

Deletion maps.  
The use of balancer chromosomes in genetic analysis.  
Using deletions to map mutations.  
Maps by somatic cell hybridization.  
Irradiated hybrid maps.  
Maps by hybridization "in situ".

#### 15. Gene expression and genetic code.

Gene expression.  
Genes coding and noncoding RNA genes.  
Characteristics of the genetic code.  
Deciphering the genetic code.  
The anticodon and the wobble hypothesis.  
Universality of the genetic code.  
Effect of codon usage.

#### 16. Molecular basis of gene mutation

How the mutation affects genetic material.  
Basic characteristics of the mutational phenomenon.  
The molecular basis of spontaneous mutation.  
Induced mutations, mutagenic chemicals and ionizing radiation

#### 17. DNA repair and recombination

A look at the repair systems.  
The molecular mechanism of recombination.  
The process of homologous recombination.  
The breakage and reunion model proposed by Holliday.  
Hybrid DNA, mismatch repair and gene conversion.

#### 18. Molecular analysis of genetic variation

DNA polymorphism concept.  
DNA polymorphism analysis of minisatellite probes.  
Analysis of DNA polymorphisms using PCR.  
The interpretation of the biological test: the fallacies of the public prosecution and defense.  
Bayesian analysis.  
Pharmacogenetics: an example of genetic individuality



### 19. Control of gene expression in prokaryotes.

Regulated and constitutive genes.  
An overview of regulation in prokaryotes.  
Global circuits of control of gene expression.  
Inducible systems: gene regulation of lactose metabolism in *E. coli*.  
Lac operon model of Jacob and Monod.  
Catabolite repression: control by positive induction in the lac operon.  
Repressible and negative control: the trp operon.  
Regulation by attenuation of the trp operon.

### 20. Control of gene expression in eukaryotes.

An overview of the levels of gene regulation in eukaryotes.  
How to activate a eukaryotic gene.  
The role of enhancers and activators of transcription factors.  
A model for activation of transcription.  
Gene regulation in the maturation of mRNA.  
Coordinated control of transcription: the role of hormones.  
Other levels of regulation.

### 21. Developmental genetics

Genetic basis of differentiation.  
Binary cell fate decisions: sex determination.  
Specification of the anteroposterior axis in *Drosophila*.  
Floral development in *A. thaliana*.  
Development and evolution processes.

### 22. Cancer genetics

Cancer as a genetic disease.  
Cell cycle control.  
Programmed cell death.  
Genetic basis of cancer: oncogenes and tumor suppressor genes.  
Hereditary predisposition to cancer.

### 23. Epigenetics

Epigenetic alterations of the genome  
Epigenetics and imprinting  
Epigenetics and Cancer  
Epigenetics and behavior  
Epigenetics and the environment



**24. Introduction to Genomics: transposable elements**

Genomics: Definition and types.  
C value paradox: genome size.  
The complexity of eukaryotic DNA.  
Genomes of prokaryotes.  
Eukaryotic nuclear genomes: genetics.  
Organelle genomes of eukaryotes  
Comparative genomics.  
The dynamic genome: transposable elements.  
Mechanisms of transposition.  
Mutagenic effects of transposition.  
Transposable elements of bacteria.  
Transposable elements of eukaryotes.

**25. Evolution of genomes**

Acquisition of new genes.  
Genomics and gene duplication.  
Duplication of domains and domain shuffling.  
Horizontal gene transfer.  
Introgression and Allopolyploidy.  
Non-coding DNA and genome evolution.  
Effect of transposable elements in the evolution of genomes.

**WORKLOAD**

ACTIVITY	Hours	% To be attended
Theory classes	57,00	100
Classroom practices	22,00	100
Laboratory practices	10,00	100
Computer classroom practice	6,00	100
Tutorials	5,00	100
Development of group work	10,00	0
Development of individual work	10,00	0
Study and independent work	50,00	0
Preparation of evaluation activities	35,00	0
Preparing lectures	25,00	0
Preparation of practical classes and problem	20,00	0
<b>TOTAL</b>	<b>250,00</b>	



## TEACHING METHODOLOGY

For the development of theoretical activities expository method and lecture are used, but by involving students with questions about cases or problems.

For practical work we use the methodology of solving exercises and problems (exercises, testing and implementation of knowledge). It strongly encourages teamwork, as both activities in the laboratory, problems, such as computer room are conducted in groups.

The activities of the course are complete and complement the transverse activity "Interdisciplinary Seminar" directly focused on the job competencies. Alternatively to this activity any other transversal activity backed by CAT can be carried out as part of a project of educational innovation

The development of the course is divided into:

### **Class work:**

A. Two or three weekly sessions of theory classes for one hour. In these sessions is present and discuss the basics of the subject with a focus on highlighting the practical aspects of the same. It is highly recommended prior reading of the issues. A total of 57 required one-hour sessions to cover this facet of teaching.

B. A weekly practical session of two hours. Here are five laboratory sessions (10 hours), eleven problem sessions (22 hours) and three practical sessions in computer lab (bioinformatics) (6 hours).

C. The availability of five sessions of one hour of tutoring group. These sessions are listed in the agenda distributed throughout the teaching period and allow us to deepen, so eminently practical and participatory on the conceptual aspects of the subject by working in groups.

### **Independent work:**

D. Interdisciplinary work: completion and presentation of a seminar. In this activity participate all subjects in the second degree course in Biology (Cell and Tissue Biology, Developmental Biology, Biochemistry, Botany, Genetics, Methods in Molecular Biology, Evolutionary Processes and Mechanisms and Zoology). The activity is mandatory for all students enrolled in second grade, except for those that have already taken it before (and have already obtained a grade). Each working group, consisting of three



students, will prepare a seminar (which consists of a written manuscript and an oral presentation) on a topic assigned at random from among those proposed by the teachers of the subjects participating in this activity. Each interdisciplinary work is considered linked (see impact assessment of the activity) to the subject that assigned the topic. A tutor will be assigned to each working team whose task will be to supervise the work to be done. To do so, there will be scheduled a series of regular meetings with the tutor throughout the course. At the beginning of the course the dates on which such meetings to follow-up the work should take place will be published, as well as the date on which the final work should be submitted and the documents handed in. A co-tutor will also be assigned who will review the final version of the work presented. Each work will be presented orally by all members of the group for 30 minutes. The presentation will be attended by all students of the course since attendance is compulsory, and also two teachers: the tutor's work and a second teacher will be attending. Both students and teachers will participate in selecting the seminars that due to their quality and originality will be presented at the Congress of Biology, in which the first and second degree courses in Biology will participate.

## EVALUATION

The assessment of learning knowledge and skills achieved by students will consider all facets of it and will essentially continuously throughout the course in order to detect possible shortcomings time student and be able to advise and help in their task. It will be therefore very important teacher-pupil relationship and knowledge on his part on the degree of learning achieved by students which will facilitated by personal tutorials.

However, in order to give a numerical rating of the degree of knowledge and skills gained by the student, will be carried out various tests that attempt to measure these from the various educational activities developed. Thus:

### A. Evaluation of the knowledge of theory.

It will assess the concepts worked in the theoretical sessions by performing two written tests and qualifying independent of matter. These tests relate to:

**A1. First test.** It will assess the theoretical, and practical application thereof, for the first 14 agenda items. Will be held during the examination period in January.

**A2. Second test.** It will assess the theoretical, and practical application of these corresponding to items 15 to 25 of the program. Will be held during the examination period of May-June.



The student is not present or does not exceed (compensable from 4 / 10) the first of these tests may recover in the period May-June reviews.

The student does not exceed the subject in the first call but must have passed (at least 5/10) of any party theory (A1 and/or A2), you keep the note of the theory adopted by the second call.

The combined value of these tests **represents 54% of the final course grade (27% each test)**.

#### **B. Assessment of knowledge and problem solving skills.**

The note of section B (problems) will be obtained from two types of activities: a) active participation in the problem classes (presentation of some of the problems that will be made during the class and realization of learning controls) and b) realization of a written test after the completion of the classroom sessions (January exam period). The student who is not present or does not pass it will be able to recover it in the examination period of May-June (first call of the subject).

**The participation grade will be worth 4 points and the written exam will be worth 16 points.** The joint value of the tests referred to this aspect represents 20% of the final grade of the subject.

To pass the subject, it is necessary to obtain a minimum grade of 4 in section B. In order to save the note in section B for the second call, it is necessary to obtain a minimum grade of 5.

#### **C. Evaluation of the work and the capacity developed during the conduct of practical laboratory work and computing.**

It will assess the work done in the lab (the labs) and practical sessions in computer lab. The assessment of this aspect will be based on the skills shown by the student to work in the laboratory or the sequence analysis program and the memory present on the work done. For laboratory work will be presented a report, while for work in computer room will be filled in questionnaires to be sent to the teacher after each session. The value of this part shall be **16 points in the final course grade (10 laboratory work and 6 computing )**.



**Attendance at lab sessions is a prerequisite to pass the course.**

#### **D. Evaluation of the interdisciplinary seminar.**

The grade obtained in interdisciplinary work will be 10% of the final grade of the course. The interdisciplinary work tutor and the professor that will attend the oral presentation will all participate in the evaluation of the this work (with a relative contribution to the activity grade of 60% and 40%, respectively). The assessment of this activity includes both the scientific content covered and presentation, and the students' ability to communicate and transmit ideas and concepts will be particularly taken into account. The papers selected for presentation at the Congress of Biology have an extra qualification, equivalent to 10% of the activity grade.

In the case the subject linked to the interdisciplinary seminar was not passed, the grade of the interdisciplinary work will be saved for the following course.

In the event that the interdisciplinary work (which is mandatory) were not carried out, the student will fail the subject linked to the interdisciplinary seminar (the subject taught by the tutor of the work who suggested the theme), regardless of the grade obtained in the rest of the parts considered for evaluation in the corresponding subject. Nevertheless if this grade were 5 or more out of 9, it will be saved for the following academic course to be added to the grade obtained in the Interdisciplinary Seminar once this is carried out carried out and passed.

If this subject were not the subject linked to the Interdisciplinary Work and this activity were not carried out, in order to pass the course students will need to obtain a grade equal to or greater than 5 on a maximum of 9.

#### **F. Student portfolio.**

The student can get up to 10% extra on the final course grade by the valuation of the interest shown by students in the subject and their degree of maturity in the field of biology, may make teachers appreciated the assistance east to the personal tutoring and individual activities that have been previously consulted with the teachers. As an example we can cite: the reading and critical analysis of books on genetics, performing certain tasks proposed for theoretical work, attendance at seminars or conferences.



Summary of the evaluation:

<b>Part</b>	<b>Punctuation</b>	<b>Retained for conv. 2 only if the note in conv. 1 is greater than or equal to :</b>
<b>A1. Lesson 1 a 14</b>	up to 27 points	<b>5</b>
<b>A2. Lesson 15 a 25</b>	up to 27 points)	<b>5</b>
<b>B. Problems</b>	up to 20 points	<b>5</b>
<b>C. Practical work (laboratory and computer science)</b>	up to 16 points (10 lab. and 6 computing)	<b>5</b>
<b>D. Interdisciplinariy seminar</b>	up to 10 points	<b>5</b>
<b>E. Student portafolio (voluntary)</b>	up to 10 points (extra)	<b>0</b>

**Other considerations:**

To pass the course will be necessary to obtain a global score above 5 on 10 (50 points) and scores greater than or equal to the equivalent of 4 out of 10 in paragraphs A1, A2, B and C. The rating of the portfolio will be taken into account once passed the course.

In the May-June exam, it will not be possible to improve the grade in sections A1 and / or B if they have already been approved (5 out of 10) in the test accomplished in January.

Laboratory and bioinformatics notes (section C, practices) or above 5 (of 10) obtained during an academic year will be saved for the calls of the three following academic years.

Students who do NOT report to any part of the final examination (theory and / or problems) and do not pass the course, include the note not present in the record.



It is recalled that it is NOT POSSIBLE TO WAIVE the qualifications above 5 obtained, both in the evaluation of the different evaluation tests and of the documents submitted for the same (exams, reports ...), as well as in the valuation of the participation in the classroom teaching activities (laboratory, problems, seminars ...).

**Second call:**

Students who fail the subject in the first round of the course (May-June), they save the note to the second round (July), either the sections on knowledge of theory independently (A1 and/or A2), the resolution problems (B), practical work (C) and / or interdisciplinary work (D), where they have passed (5 / 10). Please note that to pass the subject in the second round of the course, students must have passed the practical work (section C).

**REFERENCES****Basic**

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- Klug, W., Cummings, M.R., Spencer C. A. y Palladino M.A.(2013). Conceptos de Genética. 10ª edición. Pearson. ISBN: 9788415552499
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### Additional

- Sociedad Española de Genética (<http://www.segenetica.es/> ). Visitar el apartado de docencia: hay lecciones, problemas y recursos multimedia
- DNAi.org (DNA interactive). En inglés (<http://www.dnai.org/index.htm>)
- DNA from the beginning. En inglés. (<http://www.dnafb.org/>).
- Scitable. A Collaborative Learning Space for Science. Genetics. (<http://www.nature.com/scitable/topic/genetics-5> ).
- Departamento de Genética (<http://www.uv.es/genetica>)