

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

<b>Code</b>	33175
<b>Name</b>	Molecular genetics
<b>Cycle</b>	Grade
<b>ECTS Credits</b>	4.5
<b>Academic year</b>	2023 - 2024

**Study (s)**

<b>Degree</b>	<b>Center</b>	<b>Acad. year</b>	<b>Period</b>
1102 - Degree in Biotechnology	Faculty of Biological Sciences	3	Second term

**Subject-matter**

<b>Degree</b>	<b>Subject-matter</b>	<b>Character</b>
1102 - Degree in Biotechnology	84 - Molecular biology	Obligatory

**Coordination**

<b>Name</b>	<b>Department</b>
GIL GARCIA, ROSARIO	194 - Genetics

**SUMMARY**

Genetics is the discipline of Biology that studies the heredity and variation in living organisms. Molecular Genetics studies these processes from a chemical point of view. The focus of Molecular Genetics is the gene, its structure, organization and function.

After the identification of the chemical nature of the hereditary material in the forties of the past century, in successive stages, Molecular Genetics has been dedicated to the study of the mechanisms of the genes function and its regulation, the development of the techniques of recombinant DNA, the study of gene expression during the different stages of development and the study of the structure and composition of complete genomes. These advances have implied the interaction and interrelation of Molecular Genetics with other biological sciences, its diversification in different fields of research, and the emergence of new subdisciplines, like Genetic Engineering, Developmental Genetics and Genomics, among others.

The nature of the research and the important social repercussions of both the methods and the possible results cause a constant presence of Molecular Genetics in mass media, making it an object of debate in the most different forums. On the other hand, the power of the molecular methods and the success achieved by Genetics have also had a great impact in biological sciences, attracting towards Molecular Genetics experts from other areas like biotechnologists, medical doctors, physiologists, botanists,



microbiologists, etc., with diverse mentalities, giving rise to different questions about the expression, organization and variation of the genes.

Throughout this course, and in an attempt to complement without interferences the Genetics and Molecular Biology knowledge presented to the students in the corresponding subjects, we will focus on the study of the structure and organization of genomes throughout the evolutionary scale (virus, prokaryotes and eukaryotes; in this last case, considering both the nuclear and the organelles genomes), as well as the mechanisms involved in the dynamics and evolution of genomes. Finally, we will analyze the implication of the regulation of gene expression in the processes of differentiation and development.

## PREVIOUS KNOWLEDGE

### Relationship to other subjects of the same degree

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

### Other requirements

It is highly recommended to be enrolled or have passed the subject "Molecular Biology"

## OUTCOMES

## LEARNING OUTCOMES

### A. Conceptual aspects:

At the end of the course, the student should have acquired some basic and advanced knowledge in several aspects of Molecular Genetics, such as:

- (1) Organization of the genes in the context of the genomes in the different types of organisms.
- (2) Differences between the individual and comprehensive studies of the genes and their functions.
- (3) Genomes of model organisms and their particular interest.
- (4) Current hypotheses on the mechanisms of evolution of genomes:
  - Origin and evolution of introns.
  - Origin and evolution of virus.
  - Evolution of the genome length
  - Mechanisms of acquisition of new genes.
  - Role of transposable elements in shaping both prokaryotic and eukaryotic genomes, with special emphasis in the human genome.
- (5) Genetic bases and implications of cellular differentiation and development in multicellular organisms.
- (6) Epigenetic modifications of the genome and its importance in the phenotype

### B. Methodological aspects:

- (7) To know, understand and learn the utility, and interest of the application of genomics technologies, as well as their limitations.
- (8) To understand the importance of using combined automated sequencing methods and bioinformatics to address the study of complete genomes.



(9) To recognize the need to combine classical and molecular methodologies for the study of the genomes.

## DESCRIPTION OF CONTENTS

### 1. Basic concepts

Chemical nature and molecular structure of the genes. What is a genome. What is Genomics. Genome projects and their importance. Strategies used for sequencing and assembling complete genomes. Methods for the study of genome content: Structural and functional annotation. Comparative genomics. From genomes to cells: the transcriptome and the proteome. Metagenomics.

### 2. Molecular bases of genomes evolution

(I): Mutation and recombination. Effects of mutations: nature of the genetic code. Error-prone DNA repair: SOS response. Molecular mechanism of DNA recombination. Models to explain homologous recombination: Holliday model and Messelson-Radding modification; double-strand-break repair model. Proteins involved in the homologous recombination. Homologous recombination and repair of the DNA. Site-specific recombination.

(II): Transposition. General characteristics and classification of transposable elements. Replicative and conservative transposition: general mechanisms. Transposable elements in prokaryotes: insertion sequences, composite transposons. Transposable elements in eukaryotes: types and mechanisms. Genetic and evolutionary meaning of the transposable elements.

### 3. Genome organization in viruses

Basic features of viral genomes. Classification of viruses based on nucleic acid type. Some examples: DNA and RNA bacteriophages, DNA and RNA animal viruses. Viroids and satellite RNAs: general features. Origin and evolution of viruses.

### 4. Genome organization in prokaryotes

The concept of species in prokaryotes. Sequenced prokaryote genomes and their general characteristics. Properties of the genomes of bacteria and archaea. Length. Physical structure. Genetic organization. Introns in prokaryotes. Evolution of bacterial genomes. Impact of horizontal gene transfer. Minimal genome and Synthetic Biology. Bacterial consortia. A dynamic vision of the prokaryotic genome.



## 5. Genome organization in eukaryotes

(I): Introduction. Eukaryotes genome projects. Variation in genome length and the C-value paradox. Kinetics of reassociation and complexity of the genomes. Classification of repetitive DNA. Sequencing of model organisms. Some data from sequenced genomes of interest: *Caenorhabditis elegans*, trans-splicing and operons; the human and the chimpanzee genomes; the Neanderthal genome.

(II): Gene families. Introduction. Gene and genome duplications. Classification of homologous genes. Origin of new genes: molecular mechanisms. Families of repeated genes: rRNA genes. Families of related genes: the globins superfamily. Evolution of gene families: Mechanisms of concerted evolution.

(III): Tandem Repeat Sequences. Classification. satellite DNA: characteristics, isolation and location; origin and evolution. Minisatellite DNA: characteristics; methods of identification; DNA fingerprinting: applications. Microsatellite DNA: characteristic, methods of identification, applications. Telomeric DNA: structure, mechanism of maintenance, telomers in *Drosophila*, possible origin of telomerase.

(IV): Transposable elements. Dynamics of the transposable elements in the genome. Transposition via DNA: Classification, examples and applications. Transposition via RNA: Classification and examples. Origin of the retroelements and evolutionary relationships. Effects of the transposable elements in the genomes.

## 6. Organelles genomes

Extranuclear inheritance. General characteristics of the organelles. From endosymbionts to organelles. Structure and function of the mitochondrial genome. Mitochondrial genetic code. Characteristic of the mitochondrial DNA in fungi, plants, protozoa and animal. Structure and function of the chloroplast genome.

## 7. Genetic bases of differentiation and development

(I): Introduction. Protein profile. Totipotency, determination, differentiation and cell memory. Body plan. Embryonic territories during early development. Main decisions in the development of the embryo.

(II): Binary Decisions. Sex determination in *Drosophila*. Programmed cell death in *Caenorhabditis elegans*.

(III): Complex Decisions. Positional information: asymmetries, gradients and cellular communication. Maternal-effect genes: establishment of the polarity. Segmentation genes: compartmentalization. Homeotic genes: identity of the segments. Evolutionary conservation of the homeotic genes. Segmentation in vertebrates: somitogenesis. Influence of the environment on animal development.

(IV): Special cases of differentiation. Differentiation by DNA rearrangements: somatic recombination in the immune system. Control of cell cycle and genetic bases of cancer.





## 8. Epigenetics

Epigenetic alterations of the genome. Epigenetic and imprinting. Epigenetics and cancer. Epigenetics and behavior. Epigenetics and the environment.

## WORKLOAD

ACTIVITY	Hours	% To be attended
Theory classes	30,00	100
Classroom practices	10,00	100
Tutorials	3,00	100
Laboratory practices	2,00	100
Attendance at events and external activities	2,00	0
Development of individual work	10,00	0
Study and independent work	38,00	0
Readings supplementary material	5,00	0
Preparing lectures	12,00	0
<b>TOTAL</b>	<b>112,00</b>	

## TEACHING METHODOLOGY

### 1. Lectures:

The course will include 24 lectures of one hour. Before each lecture, a pdf file of the presentation, including relevant bibliographic references and all the essential graphic material, will be provided to the students through the “Aula Virtual” of the University of Valencia. Therefore, students can previously prepare for the session, taking only the necessary notes for their appropriate understanding.

### 2. Practical activities:

**2.1. Laboratory session.** A two hours laboratory session devoted to the “Study of mutations that affect the pattern of larvae cuticle formation in *Drosophila*”, as a case study of the process of segmentation in this organism by means of the observation of several mutants [see U. T. 7(III). Genetic analysis of development]. Attendance is obligatory. At the end of the session, the students will answer a questionnaire for its evaluation.

**2.2. Work Sessions in the classroom.** Along the course, and in accordance with the development of the theoretical sessions, the students will participate in additional activities and debates on subjects related to the studied matter. In these sessions, the active participation of the students in the discussion of the proposed subjects will be encouraged, and they will be assisted in the search of suitable material for the achievement of the posed aims.

**2.3. Critical analysis of scientific texts.** In order to promote the capacity of synthesis and analysis of the student, and his ability for proper writing communication, students will be encouraged to voluntarily present a written report including a summary and a critical discussion on a selected article proposed by



the professor and related to a subject of the program selected by the student. The deadline for the delivery of the report will be the 5 days after the associated lecture session.

### 3. Seminars:

The assistance to seminars on subjects related to Molecular Genetics with an additional interest for the students, due to the social impact of the subject or because they represent a scientific novelty, will be proposed. The speakers could be experts in the subject or a group of students. In this last case, the seminar will be exposed in the class during half an hour, and will be followed by a debate. The assistance to seminars and sessions of debate will be obligatory when they take place in the normal class schedule, and voluntary otherwise. After the seminar, the assistant students will answer a questionnaire for its evaluation, which can be voluntarily submitted in the 10 following days.

### 4. Tutorials:

The purpose of the tutorial sessions is to help and guide in a personalized manner the student, to review lecture and activities materials and address questions related to them. They also facilitate the exchange of opinions between the professor and the student. Modern technologies of information and communication can also be used for this purpose. The professor will accept queries sent by the students through the e-mail. This way will also face the student with the need to improve its written communication skills.

In addition to the individual tutorial sessions, there will be three group tutorial sessions in which questions arisen about some subject that is not well understood after the lectures or subjects that open a discussion that cannot fit in an ordinary session can be proposed by the students.

## EVALUATION

1. Evaluation of a written test: 75%. In this test the student will answer to a series of short and practical questions that represent the contents presented in the different sessions. It will be necessary to achieve at least a score of **5 over 10** in this test.
2. Evaluation of the performance on the laboratory session, by answering to a questionnaire: 5%
3. Participation in the different sessions (lectures, work sessions in the classroom, tutorials, debates and seminars) along the course: 5%
4. 15% of the global score will correspond to the participation in voluntary activities among all those proposed, being the maximum qualification for each type of activity:
  - Delivery of questionnaires relative to seminars and debates: 5%.
  - Critical analysis of scientific texts: 10%.
  - Preparation and presentation of seminars and debates: 15%.

All activities will be processed through Aula virtual. Detailed instructions and evaluation criteria for each activity are available in the folder "Resources". All documents relative to the activities must be delivered through the "Tasks" application. The scores will appear in each Student card.

To apply for advanced evaluation, the student must have done before the mandatory activities specified in this Course Guide.



## REFERENCES

### Basic

- Brown, T.A. (2007). Genomes 3 (3rd edition). Oxford. (Traducido al castellano en Editorial Medica Panamericana).
- Pierce, B. A. (2014). Genetics : A conceptual approach, 5th edition. W. H. Freeman and Co. (Traducido al castellano en 2016 por Editorial Médica Panamericana).
- Krebs, J.E., E.S. Goldstein and S.T. Kilpatrick (2014). Lewin's Genes XI. Jones and Barlett Publishers. (Genes IX está traducido al castellano en 2008 por McGrawHill Interamericana Editores).
- Hartl, D.L. y Jones, E.W. (2005). Genetics: Analysis of genes and genomes, 6th edition. Jones and Bartlett.
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- Klug, W. S. et al. (2012). Concepts of Genetics (10th edition). Pearson Education. (Traducido al castellano en 2013).
- Benito, C. y Espino F.J. (2013). Genética. Conceptos esenciales . Editorial Médica Panamericana.

### Additional

- Clark, M. (2000). Comparative Genomics. Kluwer Academic Publishers
- DOE Joint Genome Institut. Why sequence them? <http://www.jgi.doe.gov/sequencing/why/index.html>
- Fontdevila, A. y Moya, A. (2003). Evolución. Origen, adaptación y divergencia de las especies. Editorial Síntesis.
- Gilbert, S. F. (2006). Developmental Biology, 8th edition. Sinauer Associates Inc. Publishers. (Traducido al castellano en Editorial Medica Panamericana)
- Gregory, T. R. (2006). The evolution of the genome. Elsevier.
- Hartwell, L., Hood, L., Goldberg, M.L., Silver, M.L., Veres, R.C. y Reynolds, A. (2004). Genetics: from genes to genomes, 2nd edition. McGraw-Hill.
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