

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

<b>Code</b>	33137
<b>Name</b>	Genomics
<b>Cycle</b>	Grade
<b>ECTS Credits</b>	4.5
<b>Academic year</b>	2022 - 2023

**Study (s)**

<b>Degree</b>	<b>Center</b>	<b>Acad. year</b>	<b>Period</b>
1109 - Degree in Biochemistry and Biomedical Sciences	Faculty of Biological Sciences	3	First term

**Subject-matter**

<b>Degree</b>	<b>Subject-matter</b>	<b>Character</b>
1109 - Degree in Biochemistry and Biomedical Sciences	9 - Genética y biología molecular	Obligatory

**Coordination**

<b>Name</b>	<b>Department</b>
PARICIO ORTIZ, NURIA	194 - Genetics

**SUMMARY**

Genetics is the part of Biology that deals with the study of heredity and variation in organisms. Molecular Genetics studies these processes from the chemical point of view. Genomics is the analysis of the genome after sequencing, with particular emphasis on comparative aspects related to their composition and the factors that promote its evolution. This is a still young biology field but it has growing interest from the development of new technologies that allow the characterization of more genomes, and thus open the possibility of explaining their structures, functions, and according to their compositions, their evolution. It is, therefore, a field which provides explanations to classic questions of biology: the origin of new functions, evolution of complexity, speciation, the role of non-coding DNA, the increase in genome size, etc..

After identifying the chemical nature of hereditary material in the mid-40's last century, Molecular Genetics and more recently Genomics are dedicated to the study of mechanisms of action and regulation of genes, the development of recombinant DNA techniques, the study of gene expression during different stages of development and the study of the structure and composition of whole genomes. These advances have led to the interaction and interrelationship of Molecular Genetics with other sciences, generating new subdisciplines, such as Genetic Engineering, Molecular Evolution, Genetics of Development,



EvoDevo, and, more recently, Genomics.

The nature of research and the enormous social implications of the methods as well as the eventual results, makes that Genomics has a constant presence in the media and is the subject of debate in the most diverse forums. Moreover, the power of molecular methods and the success of genetics has been felt in other areas of biology, pulling experts from other areas such as biotech scientists, physicians, physiologists, botanists, microbiologists to Molecular Genetics and Genomics, which raise different issues on topics such as expression, organization and variation of genes.

## 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

### 1101 - Degree in Biochemistry and Biomedical Sciences

- Skills in analysis and synthesis.
- Capacidad de resolución de problemas.
- Capacidad de aprendizaje autónomo.
- Capacidad de comunicación oral y escrita.
- Capacidad de manejar el inglés como lengua extranjera.
- Capacidad de utilizar las nuevas tecnologías de información y comunicación.
- Desarrollo de habilidades para comprender metodología e interpretar resultados científicos.
- Capacidad para el trabajo en equipo y la cooperación.
- Desarrollo de la capacidad de razonar y aplicar el método científico.
- Comprensión de la lógica molecular de los seres vivos como producto de la evolución.
- Conocer y comprender las bases moleculares de la información genética y los mecanismos de su transmisión y variación.
- Relacionar las características estructurales y funcionales de las macromoléculas.
- Tener una visión integrada del metabolismo celular y la expresión génica relacionándolas con los distintos compartimentos celulares.
- Conocer los elementos comunes y los diversos de la genética y la biología molecular de los diferentes tipos de organismos vivos.



- Adquirir conocimientos teóricos sobre la estructura, función y evolución de los genomas.
- Conocer las aplicaciones de los conocimientos en genética y biología molecular en el diagnóstico de enfermedades humanas.

## LEARNING OUTCOMES

### Conceptual Aspects

It aims to extend the knowledge acquired in the Genetics subject, related to Molecular Genetics and genomics. Specifically, it is the student acquire the basic knowledge about the following:

- (1) Gene organization and function in the context of their genomes in different types of organisms.
- (2) Methods of analyses and characterization of genomes, both in prokaryotes and eukaryotes. Metagenomes.
- (3) Genomes of model organisms and their particular interest. Characteristics of the human genome.
- (4) Current hypotheses on the mechanisms of genome evolution:
  - Evolution of genome size
  - Mechanisms for acquisition of new genes.
  - Role of transposable elements in both prokaryotes and eukaryotes, with particular emphasis on studying the human genome.
  - Mutations and DNA repair.
  - Molecular aspects of recombination.
  - Origin of genomic complexity.

### Methodological aspects

- (5) To know, understand and learn the value and interest to the application of genomic technologies, as well as its limitations:
- (6) To understand the importance of combined use of automated sequencing methods and bioinformatics techniques to address the sequencing of entire genomes.
- (7) To understand the need to combine classical and molecular methodologies for the study of genomes.



The student should be familiar with the following contents:

Analysis, structure and organization of genomes. Genomic study of communities of microorganisms, metagenomics. Functional analyses of genomics: transcriptomics and proteomics. Genomic size and complexity. Role of horizontal transfer in prokaryotes. Increasing genomic complexity. Origin of new genes in eukaryotes. Importance of gene duplication and / or genomics: 2R hypothesis. Role of non-coding DNA in genomes. Mechanisms of evolutionary change. Role of transposable elements. Mechanisms of transposition. DNA recombination.

## DESCRIPTION OF CONTENTS

### 1. Basic concepts in Genomics (I).

What is a genome. What is genomics. Fields in genomics. Genome projects and their importance. Strategies used for sequencing and assembly of complete genomes. Determination of gene localization and function: computational analysis and experimental techniques.

### 2. Basic concepts in Genomics (II).

From genomes to cells: the transcriptome and proteome. Variations in the transcriptome in eukaryotes: alternative splicing and RNA editing. Role of introns in the evolution of genomes. Two theories about their origin. Variations in the transcriptome and proteome in eukaryotes. Other genomic analyses.

### 3. Organization of viral genomes.

Basic features of the genome of the virus. Classification according to their hereditary material. Examples: RNA and DNA bacteriophages, DNA and RNA animal viruses. Viroids and satellite RNAs: general features. Origin and evolution of viruses.

### 4. Genome organization in prokaryotes.

Generalities. Basic features of the prokaryotic genome: size, physical structure, genetic organization and content. Bacterial transposable elements: insertion sequences, compound transposons. The species concept in prokaryotes. Evolution of the prokaryotic genome: the role of horizontal gene transfer and reductive evolution.

### 5. Peculiarities of archaea.

Basic concepts. Features of archaea. Identification of new archaea: molecular methods. Classes of archaea. Comparison of archaea to eukaryotes and bacteria. Role of archaea in the origin of the eukaryotic cell.



## **6. Genome organization in eukaryotes (I).**

Introduction. Variation in genome size and the C value paradox Reassociation kinetics and genome complexity. Classification of repetitive DNA: functional sequences and non-functional sequences. Some data from sequenced genomes: *Caenorhabditis elegans*, operons and "trans-splicing". The human genome: associated projects.

## **7. Genome organization in eukaryotes (II).**

Gene families: types. Gene and genomic duplication. Origin of new genes: molecular mechanisms. The 2R hypothesis. Families of repeated genes: rDNA, tRNA, histones. Families of related genes: the globin cluster. Paradox of the gene families. Concerted evolution.

## **8. Genome organization in eukaryotes (III).**

Tandemly repeated sequences. Main classes in human DNA. Satellite DNA: characteristics, location and function. Minisatellite and microsatellite DNA: characteristics and applications: DNA "fingerprinting". Telomeric DNA: structure. Mechanisms of telomeres maintenance.

## **9. Genome organization in eukaryotes (IV).**

Transposable elements. Classification of transposable elements. DNA transposons: Control elements in maize, P elements in *Drosophila melanogaster*, and other elements. Retrotransposons: Retroviruses, retrotransposons with LTRs, retrotransposons without LTRs. Transposable elements in the human genome. Effects of transposable elements in the genome.

## **10. Organelle genomes.**

Extranuclear inheritance. General features of the organelles. From endosymbionts to organelles. Structure and function of the mitochondrial genome. Mitochondrial genetic code. Characteristics of mitochondrial DNA in yeast, animals and plants. Structure and function of the chloroplast genome.

## **11. Molecular basis of genomic evolution (I): Mutation and repair.**

Causes of mutation. Types of mutations. Effects of mutations. Reversion. Spontaneous and induced mutations. Reversion. Error prevention. DNA repair: mechanisms. Direct repair: types and genes involved. Excision repair: types and genes involved. Recombination repair.

**12. Molecular basis of genomic evolution (II): Recombination.**

Types of recombination. Models of homologous recombination: Holliday model and heteroduplex DNA, Meselson-Radding model, double-stranded break model. Gene conversion. Proteins involved in homologous recombination. Site-specific recombination. Somatic recombination and hypermutation: immunoglobulin genes.

**13. Molecular basis of genomic evolution (III): Transposition.**

General mechanisms of transposition: replicative and conservative transposition. Genetic and evolutionary significance of transposable elements.

**WORKLOAD**

ACTIVITY	Hours	% To be attended
Theory classes	36,00	100
Classroom practices	6,00	100
Tutorials	3,00	100
Attendance at events and external activities	2,50	0
Development of group work	10,00	0
Development of individual work	10,00	0
Study and independent work	10,00	0
Readings supplementary material	5,00	0
Preparation of evaluation activities	5,00	0
Preparing lectures	25,00	0
<b>TOTAL</b>	<b>112,50</b>	

**TEACHING METHODOLOGY**

The development of the subject is structured around theoretical and working sessions in the classroom, computer sessions, tutoring in person or via e-mail, preparation of presentations and reports individually and in groups and attendance at seminars, taught by experts or by the students themselves.

- Theoretical sessions:** In the section on classroom work, 28 sessions of one-hour are included. Before starting each lesson, students will have a script of it, which will include the theoretical content of the sessions, all significant artwork that will be presented and a section with the latest bibliographic contributions on the subject. This script will be available in the Virtual Classroom of the University of Valencia. Thus, it is intended that the student can prepare classes in advance that will be easily followed, only taking the notes needed for proper understanding.
- Working sessions in the classroom:** Throughout the course, and in accordance with the development of theoretical sessions, it will be offered to students the possibility to participate in



debates/seminars on issues related to the subject studied. To this end, working groups of 2-3 students will be established for the preparation of such debates/seminars under the guidance of the teacher, who will assist them in finding suitable material to achieve the objectives. In these meetings, the active participation of students in the discussion of the proposed item will promote and, upon completion, students will have to elaborate a summary on the topic under discussion. In addition, several sessions will be held in the computer lab in which programs for genome analysis and databases related to the human genome available in free websites will be used. For better utilization of this activity, it will be raised for subgroups of 16 students.

3. **Seminars, conferences and other activities:** They will serve to develop activities that will allow students to expand their knowledge on the subject and to relate it with other disciplines, and to promote the acquisition of skills different than those acquired in the lectures and work sessions classroom. One of these activities will be the critical analysis of scientific papers selected by the teacher of the subject. This activity aims at training the student in the reading of scientific papers (which necessarily involves reading technical English), bringing him/her closer to the original literature that permits the development and advancement of biomedical sciences. This activity is mandatory, and it will be organized jointly with the other subjects in the same course. The preparation, presentation and discussion (30 minutes) of the scientific papers will be held in groups of 2 students and supervised by the teacher through the tutorials. The attendance to seminars and conferences on topics related to genomics will be also promoted, specially those that may present an additional interest to students, as may be the social impact of the topic or the presentation of some high-profile scientific novelty. These seminars will be conducted by experts on the subject. In order to develop the critical ability of the students, they may voluntarily submit a report on the seminar or conference received, consisting of a brief summary on it and a critical discussion.
4. **Tutorials:** The role of tutoring is to help and guide the student personally in all the problems that arise in dealing with the study of the subject. They facilitate the exchange of views between teacher and student, in an effort to individual approach to teaching. The information and communication technology can also be used to enhance teacher-student interaction. Queries sent by students via e-mail will be accepted, configuring a remote tutoring. Since they are a way of written communication, the student is forced to perform an analysis of their own doubts and to learn to clearly express them in writing. In addition to personal tutorials, we propose to hold three group tutoring sessions in which the teacher and the students will jointly resolve issues that have not been sufficiently clarified during regular meetings or those issues that gave rise to further discussion and that would not fit in regular sessions. For better utilization of these activities, they will be raised for subgroups of 16 students.

## EVALUATION

- Evaluation of the knowledge acquired during the lectures will be made by conducting a written test in which the student must answer a series of short questions and practical issues covering all the contents in class. The note of this test represent 75% of the total grade. To pass the course, a score of 5 out of 10 in that test should be reached.
- The remaining 25% of the grade can be obtained through participation in various activities to achieve the maximum allowed. The maximum score for each section is as follows:



- Presentations of topics related to the subject and delivery of reports about them: 10%.
- Solving of questionnaires in tutoring sessions: 10%.
- Critical analysis of scientific papers: 5%.

To evaluate the activity of critical analysis of articles, the following evaluation criteria will be also considered: knowledge and understanding of the information contained in the articles, correct use of terminology and speaking skills. Integration with other theoretical and practical content of this or other subjects of the grade will be also valued. The maximum score in this activity will be of 10 points, being needed 5 points to overcome it. The mark obtained will represent 5% of the final grade for each of the subjects in the third year that participate in this activity. If the student does not reach the minimum score required, suspend the subject in which he/she prepares this activity. The participation of other students in the presentation and the discussion sessions may be also taken into account by the teacher to modulate the final mark of the subject.

## REFERENCES

### Basic

- Alberts B., Johnson A., Lewis J., Morgan D., Raff M., Roberts K. y Walker P. (2014). Molecular Biology of the Cell, 6th edition. Garland Science Editions.
- Brown T. A. (2017). Genomes 4, 4th edition. Garland Science. (3ª edición traducida al castellano en Editorial Médica Panamericana).
- Hartwell L., Goldberg M., Fischer J. y Hood L. (2018). Genetics: from genes to genomes, 6th edition. McGraw-Hill.
- Krebs J. E., Goldstein E. S. y Kilpatrick S. T. (2017). Lewins Genes XII. Jones & Barlett Publishers.
- Lesk A. M. (2017). Introduction to Genomics, 3rd edition. Oxford University Press.
- Pierce B. A. (2020). Genetics: A conceptual approach, 7th edition (3ª edición traducida al castellano). Mcmillan Learning.
- Strachan T. y Read A. (2018). Human Molecular Genetics, 5th Edition. CRC Press, Taylor & Francis Group.
- Watson J. D., Baker T. A., Bell S. P., Gann A., Levine M. y Losick R. (2014). Molecular Biology of the Gene, 7th edition. Pearson Education, Inc.
- Ginsburg G y Willard H. (2013). Genomic and personalized medicine, 2nd Edition. Elsevier.
- Klug W. S., Cummings M. R., Spencer C. A. y Palladino M. A. (2019). Concepts of Genetics, 12th edition Pearson Education, Inc. (10th edición traducida al castellano).

### Additional

- BacMap: An Interactive Atlas for Exploring Bacterial Genomes: <http://bacmap.wishartlab.com/>
- DOE Joint Genome Institute: <http://genome.jgi.doe.gov/>



- GOLD (Genomes Online Database). <http://www.genomesonline.org/>
- Human Genome Project Information :  
[http://web.ornl.gov/sci/techresources/Human\\_Genome/index.shtml](http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml)
- KEGG (Kyoto Encyclopedia of Genes and Genomes) Genome database:  
<http://www.genome.jp/kegg/genome.html>
- The Encyclopedia of DNA elements (ENCODE) Consortium: <http://genome.ucsc.edu/ENCODE/>
- The modENCODE project: <http://www.modencode.org/>
- Base de datos del genoma humano: [http://www.ensembl.org/Homo\\_sapiens/Info/Index](http://www.ensembl.org/Homo_sapiens/Info/Index)
- The Roadmap Epigenomics Project: <http://www.roadmapepigenomics.org/>
- Sitios web relacionados con la enseñanza en Genómica (NIH): <https://www.genome.gov/about-genomics/teaching-tools/Genomics-Education-Websites>