

**COURSE DATA****DATA SUBJECT****Code:** 33155**Name:** Human genetics**Cycle:** Undergraduate Studies**ECTS Credits:** 6**Academic year:** 2025-26**STUDY (S)**

Degree	Center	Acad. year	Period
1109 - Degree in Biochemistry and Biomedical Sciences	Facultat de Ciències Biològiques	4	First quarter

**SUBJECT-MATTER**

Degree	Subject-matter	Character
1109 - Degree in Biochemistry and Biomedical Sciences	Biomedicina molecular	COMPULSORY

**COORDINATION**

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**SUMMARY**

Human Genetics is a subject belonging to the fourth year of the Degree in Biochemistry and Biomedical Sciences (University of Valencia, Plan 2009). It is a theoretical and practical subject of the first semester of the course, containing 6 ECTS credits. It is part of the subject "Molecular Biomedicine" along with "Clinical Biochemistry and Molecular Pathology", and the optional subjects "Microbial Pathogenesis", "Molecular Pharmacology" and "Molecular and Public Health Parasitology".

Human genetics studies heredity and variation of biological traits in human, being one of the cornerstones of Biomedicine. Knowledge of the language, concepts and methods of Human genetics and the assessment of genetic and genomic perspective of health and disease, provide a learning framework which is essential in the professional practice within the Health Sciences and Biomedicine at the present time.

This subject provides a comprehensive and integrated application of genetics to the study of heredity in human beings, with emphasis on specific methods and techniques that have allowed the isolation of genes and characterization of mutations causing many hereditary diseases. In keeping with the theoretical development of the subject, will be experimentally developed some of the methods used currently in the implementation of Genetic counseling and Genetic diagnosis in its different facets, complemented with the discussion and resolution of specific clinical cases.



## PREVIOUS KNOWLEDGE

### RELATIONSHIP TO OTHER SUBJECTS OF THE SAME DEGREE

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

### OTHER REQUIREMENTS

## COMPETENCES / LEARNING OUTCOMES

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Acquire skills to use the methodologies of molecular biosciences and to keep an annotated record of activities.

Be able to use new information and communication technologies.

Capacidad para el trabajo multidisciplinar en equipo y la cooperación.

Capacidad para trabajar correctamente en los laboratorios de Biomedicina incluyendo seguridad, manipulación, eliminación de residuos y registro anotado de actividades.

Capacidad para utilizar la instrumentación básica en los laboratorios de Biomedicina.

Conocer el papel de los genes en el cáncer y su seguimiento mediante marcadores tumorales.

Conocer las aplicaciones de los análisis genéticos en la identificación de individuos y la determinación de relaciones de parentesco.

Conocer las estrategias genéticas para la prevención de enfermedades hereditarias como son el consejo y el diagnóstico genético.

Conocer los fundamentos de terapia génica.

Conocer los principales métodos y técnicas experimentales aplicadas al estudio de la salud y enfermedad humanas, su etiología y la efectividad de los tratamientos.

Conocimiento de las enfermedades y disfunciones más frecuentes.

Desarrollo de habilidades para la aplicación de los conocimientos adquiridos al mundo profesional.

Diferenciar entre enfermedades cromosómicas, de transmisión mendeliana y multifactoriales.

Have capacity for analysis, synthesis and critical reasoning in the application of the scientific method.

Know and understand the molecular bases of genetic information and the mechanisms of its transmission and variation.

Know how to apply the knowledge gained in the diagnosis, prevention and treatment of human diseases.



Know how to design multidisciplinary experimental strategies in the field of molecular biosciences to solve complex biological problems, especially those related to human health.

Know how to use the different bibliographic sources and biological databases and be able to use bioinformatic tools.

Know how to work responsibly and rigorously in the laboratory, considering the safety aspects in experimentation as well as the legal and practical aspects of the handling and disposal of waste.

Know the common and differential molecular and cellular elements of the different types of living organisms with special emphasis on the human being and model organisms for their study.

Know the usual procedures used by scientists in the area of molecular biosciences and biomedicine to generate, transmit and disseminate scientific information.

Tener una visión integrada de las técnicas y métodos utilizados por las ciencias Biomédicas.

Understand experimental approaches and their limitations and interpret scientific results in molecular biosciences and biomedicine.

Utilización de terminología específica de la biomedicina.

## DESCRIPTION OF CONTENTS

### 1. Introduction to human Genetics

The role of Human Genetics in Biomedicine and Health Sciences. Technical and scientific advances in Human Genetics. The Human Genome Project. Major international projects in genomics. Genomic databases. Medical genetics. Genetic diseases and hereditary diseases. Prevalence and Incidence of a disease.

### 2. The genetic and molecular basis of Mendelian inheritance patterns.

Rare diseases. Mendelian inheritance. Gain and loss of function mutations. Genealogy studies. Inbreeding coefficient. Autosomal and sex chromosome-linked inheritance patterns. Examples. Atypical patterns of inheritance. Multiple alleles: Blood groups and HLA antigens.

### 3. Heterogeneity and variable expression of Mendelian phenotypes.

Factors affecting basic patterns of inheritance. Penetrance. Expressivity. De novo mutations. Gonadal mosaicism. Lethality. Genetic heterogeneity. Pleiotropy.



#### **4. Non-classical Mendelian inheritance: Dynamic mutations and genomic imprinting.**

Dynamic mutations. Mechanisms and classification of nucleotide expansions. Genetic anticipation. Fragile sites. Epigenetics. Epimutations. Epigenetic and epigenomic diseases. Imprinted genes.

#### **5. Mitochondrial inheritance**

The human mitochondrial genome. Characteristics of mitochondrial inheritance. Types of mutations in the mitochondrial genome. Diseases with mitochondrial inheritance. Examples. Mitochondrial replacement therapy.

#### **6. Genetic diseases with complex inheritance**

Genes and Environment. The norm of reaction. Continuous multifactorial inheritance. Discontinuous multifactorial inheritance. Alleles of genetic vulnerability. Study of complex diseases: studies of family clustering; studies of twins: rates of concordance and heritability; adoption studies.

#### **7. Identifying Genes Causing Mendelian Diseases**

Principles and strategies. Conventional methodologies: functional cloning, candidate gene approaches, and positional cloning. Genetic mapping techniques: cloning chromosomal breakpoints, linkage analysis, and autozygosity mapping. Genomic methodologies: whole genome/exome sequencing. Decisions and criteria for identifying the causal gene. Gene panels. Ethical issues: incidental findings, secondary findings, and opportunistic screening.

#### **8. Identifying Genetic Risk Factors in Complex Inheritance Diseases**

Introduction. Case-control association studies. Linkage disequilibrium. Genome-wide association studies (GWAS). Polygenic risk. Limitations of association studies. Gene-environment interaction (GxE) in complex diseases.



## **9. Cancer Genetics**

Introduction. Genetic and environmental risk factors. Somatic mutations. Germline mutations. Oncogenes and activation mechanisms. Tumor suppressor genes. Knudson's hypothesis. Tumoral loss of heterozygosity. DNA repair alterations. Chromosomal instability. Tumor progression. Examples.

## **10. Genetic Counselling**

Genetic counseling. Determining recurrence risks. Genetic diagnosis: preimplantation, prenatal and presymptomatic. Diagnostics and laboratory tests. Population genetic screening.

## **11. Genetic diseases treatment**

Treatment strategies. Gene therapy: types. Therapeutic genes and constructs. Transfer vectors and methods. Candidate diseases. Applying genomics to individualize cancer therapy. Pharmacogenetics and personalized medicine. Ethical issues in human genetics.

## **12. Human Genetics Laboratory: Detection of polymorphisms in the ADH3 gene using filter hybridization and ASO probes.**

Isolation gDNA from peripheral blood samples. Amplification of ADH3 exon 8 by PCR from gDNA. Dot blot. Hybridization with dUTP-digoxigenin-labeled allele-specific oligonucleotides (ASO probes). Detection of hybrids by colorimetry. Analysis of results.

## **13. Human Genetics Laboratory: Karyotyping**

Extraction of 2 ml of peripheral blood. Lymphocyte culture. Accumulation of cells in metaphase and dispersion of chromosomes. Fixation, extension and staining of the samples. Observed under the optical microscope and photographed. Ordering and classification of chromosomes.

## **WORKLOAD**

**PRESENCIAL ACTIVITIES**

Activity	Hours
Theory	36,00
Laboratory	16,00
Classroom practices	8,00
<b>Total hours</b>	<b>60,00</b>

**NON PRESENCIAL ACTIVITIES**

Activity	Hours
Attendance at other activities	1,00
Individual or group project	6,00
Independent study and work	40,00
Preparation of lessons	10,00
Preparation for assessment activities	19,00
Resolution of case studies	14,00
<b>Total hours</b>	<b>90,00</b>

**TEACHING METHODOLOGY**

**1. Lecture Classes.** The lecture classes will primarily consist of traditional lectures lasting one hour. The instructor will present the most relevant content of the course, using the necessary audiovisual aids to ensure dynamic and coherent delivery. The instructor will make the necessary materials available on the Virtual Classroom teaching platform, allowing students to effectively follow the classes. The content will be integrated with that of the other course activities, while also promoting cross-disciplinary connections with other subjects within the "Molecular Biomedicine" area.

**2. Laboratory Classes.** The laboratory practical program will be developed in coordination with the lecture classes. The duration of each laboratory session will be between 2 and 4 hours depending on the day. Attendance at these sessions is mandatory.

**3. Problem-Solving Classes.** These 1-hour sessions will reinforce the concepts presented during the lectures and encourage active student participation through the discussion and resolution of problems and clinical cases. The instructor will prepare a set of exercises for each thematic block, allowing students to work both individually (through personal preparation) and collectively (through presentation and discussion in group classes) on the most relevant topics.

**4. Seminars, Guest Lectures, or Other Activities.** Seminars will be delivered by researchers whose lines of work are related to Molecular Biomedicine. These guest speakers will provide students with the opportunity to learn about and engage with current research in areas relevant to their studies.

**5. Personalized Tutorials.** Students will be encouraged to make use of this resource to seek guidance and discuss with the instructor any topic related to the course, or their undergraduate studies.

**EVALUATION**



- **Objective tests on the contents of the subject (80%):** (i) The evaluation of theoretical knowledge will constitute 60% of the final grade. This will be assessed through multiple-choice tests and written exams with short-answer questions; (ii) Additionally, 20% of the final grade will be based on the student's ability to address and solve human genetics problems. This will be evaluated through a written test administered alongside the theoretical exam at the end of the course.

- **Individualized follow-up in practical activities (20%):** (i) Assessment will include the presentation and discussion of laboratory results, which will take place 15 days after the conclusion of these classes (10% of the final grade), and (ii) a written test (10% of the final grade) to be conducted during the last laboratory session. Attendance at the lab sessions is a prerequisite for passing the course.

The final grade will be the sum of the marks achieved in the different sections. To pass the course, a cumulative score of 5 out of 10 or higher is required, provided that both theoretical and practical knowledge (including problem-solving and laboratory work) independently achieve a minimum of 4 out of 10.

For students who do not pass the course on the first attempt, marks for theoretical knowledge, problem-solving, and laboratory will be carried over to the second attempt if each mark is 4 out of 10 or higher.

The laboratory mark will be carried over to the next academic year if it is 5 out of 10 or higher.

Students who do not attend any part of the final exam (theory or problems) will receive a 'NOT EVALUATED' status in the records.

To request an early examination date for this course, all mandatory activities outlined in this teaching guide must be completed

## REFERENCES

### Basic Bibliography

- COHN R, SCHERER S and HAMOSH A. Thompson & Thompson Genetics and Genomics in Medicine. 2023 (9<sup>a</sup> Ed). Editorial Elsevier. ISBN: 978-0323547628.

- JORDE LB, CAREY JC y Bamshad MJ. Genética Médica. 2021 (6<sup>a</sup> ed). Editorial Elsevier España SL. ISBN 978-84-9113-797-9; eISBN: 978-84-9113-880-8.

- PIERCE B.A. Genetics: A Conceptual Approach. 2018 (5<sup>a</sup> Ed.). WH Freeman. ISBN 1319187811. La cuarta edición tiene traducción al castellano: Genética: un enfoque conceptual. 2016 (4<sup>a</sup> ed). Editorial Médica Panamericana.

- PRITCHARD D.J. / KORF B.R. Genética Médica. 2015 (3<sup>a</sup> ed). Editorial Médica Panamericana. ISBN 9788479033958

- STRACHAN T, GOODSHIP J, and CHINNERY P. 2015. Genetics and Genomics in Medicine. Garland Science, Taylor & Francis Group, LLC. ISBN 978-0-8153-4480-3.



- STRACHAN and READ. Human Molecular Genetics. 2019 (5<sup>a</sup> ed, ISBN 0815345895); 2010 (4<sup>a</sup> ed, ISBN 9780815341499) Garland Science/Taylor & Francis Group. La tercera edición tiene traducción al castellano (2006, Mc. Graw-Hill Interamericana, ISBN970-10-5135-1).

-TURNPENNY P. and ELLARD S. Emery. Elementos de genética médica. 2018 (Ed. 15). Elsevier. ISBN 9788491132066.

### **Supplementary Bibliography**

- DELGADO RUBIO A., GALÁN GÓMEZ E., GUILLÉN NAVARRO E., LAPUNZINA BADÍA PABLO D., PENCHASZADEH VICTOR B., ROMEO CASABONA CARLOS MARÍA, EMALDI CIRIÓN AITZIBER. Asesoramiento Genético en la práctica médica. 2012. Editorial Médica Panamericana.

- Recommended websites:

- GeneCards: <http://www.genecards.org/>
- Gene Names: [www.genenames.org](http://www.genenames.org)
- GeneReviews: <http://www.genereviews.org/>
- OMIM - Online Mendelian Inheritance in Man: <https://omim.org/>
- Orphanet (portal europeo de información de referencia en enfermedades raras y medicamentos huérfanos): <https://www.orpha.net/es>