

**COURSE DATA****DATA SUBJECT****Code:** 33155**Name:** Human genetics**Cycle:** Undergraduate Studies**ECTS Credits:** 6**Academic year:** 2026-27**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 compulsory fourth-year course in the Bachelor's Degree in Biochemistry and Biomedical Sciences at the Universitat de València. It comprises both theoretical and practical components and is taught during the first semester of the academic year. The course is part of the Molecular Biomedicine module, together with the compulsory course Clinical Biochemistry and Molecular Pathology, and the elective courses Microbial Pathogenesis, Molecular Pharmacology, and Molecular Medical Parasitology.

Human Genetics examines the mechanisms of inheritance and genetic variation in humans and constitutes a fundamental discipline for understanding the molecular basis of health and disease. The aim of this course is to provide a comprehensive and integrated overview of the application of genetics to the study of human inheritance, covering both classical principles and the latest advances in molecular genetics, genomics, and epigenetics.

The course also addresses the methods and techniques that have enabled the identification and characterization of genes involved in hereditary disorders, as well as genetic variants associated with susceptibility to complex multifactorial diseases. In addition, it examines the main strategies for the prevention of genetic diseases, which are based on three fundamental pillars: genetic counselling, genetic



diagnosis, and population-based genetic screening programmes.

The theoretical content is reinforced through the discussion and resolution of problem sets and case studies based on real clinical scenarios, as well as through the experimental application of fundamental methodologies for chromosome analysis, the study of genetic variation, and the interpretation of molecular data. Ultimately, the course provides students with the conceptual and methodological foundations required to critically interpret genetic information, use the terminology of Human Genetics accurately, and understand the impact of advances in genomics and personalized medicine on contemporary biomedical practice.

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

1101 -

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. Postzygotic 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

Fundamental characteristics of cancer. Types of cancer and incidence. Genetic and environmental risk factors. Somatic mutations. Germline mutations. Hereditary predisposition to cancer. Oncogenes and activation mechanisms. Tumor suppressor genes. Knudson's hypothesis. Loss of tumor heterozygosity. Alterations in DNA repair. 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). Deteccion 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
Total hours	60,00

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
Total hours	90,00

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 sessions. Laboratory classes will be conducted in five sessions lasting between 2 and 4 hours, depending on the day (16 hours in total), and attendance is mandatory. Changes between laboratory groups will only be permitted through a mutual exchange between students before the start of the laboratory sessions or when places are available in another group.

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.

- **Continuous assessment of practical activities (20%):**

The acquisition and understanding of the content covered during the laboratory sessions will be assessed through a multiple-choice test administered during the final laboratory session. This assessment will account for 10% of the final course grade.

Students' attitude, participation, and engagement during the laboratory sessions, together with the preparation and submission of a laboratory report presenting the results obtained, as well as their discussion and interpretation, will account for the remaining 10% of the final course grade. The laboratory report must be submitted within 15 calendar days after the completion of the laboratory sessions.

The laboratory activities are non-recoverable, and students must attend at least 80% of the laboratory sessions to be eligible to submit the laboratory report. Any absence must be duly justified.

The final grade for the first examination period will be the sum of the marks obtained in the different assessment components. To pass the course, students must obtain an overall grade of at least 5.0 out of 10, provided that the mark obtained in each of the three components (theory, problem-solving, and laboratory) is at least 4.0 out of 10.

Marks obtained in each assessment component (theory, problem-solving, and laboratory) during the first examination period will be carried forward to the second examination period, provided that the mark for the corresponding component is 4.0 out of 10 or higher.

The final grade for the second examination period will be calculated according to the same criteria as for the first examination period, except in cases where the student has attended less than 80% of the laboratory sessions. In such cases, students will be required to take a laboratory examination covering the



theoretical concepts and the rationale of the laboratory methodologies used. This examination will consist of short-answer and multiple-choice questions and will be held on the date scheduled for the second examination period. Since no laboratory report will have been submitted, the maximum final grade attainable in the course will be 9.0 out of 10. As in the first examination period, students must obtain an overall grade of at least 5.0 out of 10, provided that the mark in each assessment component (theory, problem-solving, and laboratory examination) is at least 4.0 out of 10.

Laboratory marks of 5.0 out of 10 or higher will be carried forward to the following academic year.

Students who do not attend any of the assessment components, either in the first or the second examination period, will be recorded as Absent (No Presentado) in the official records. Likewise, if the sum of the marks obtained in the three assessment components is 5.0 out of 10 or higher, but one of the components has a non-compensable mark (i.e., below 4.0 out of 10), the final grade recorded in the official records will be Fail. In this case, the compensable marks obtained in the remaining assessment components will be carried forward to the second examination period.

REFERENCES

Basic Bibliography

- COHN R, SCHERER S and HAMOSH A. Thompson & Thompson Genetics and Genomics in Medicine. 2023 (9^a Ed). Editorial Elsevier. ISBN: 978-0323547628. A Spanish translation is available. (Genética y genómica en medicina, 2024 (9^a Ed). Editorial Elsevier. ISBN: 9788413826974).
- JORDE LYNN B., CAREY JOHN C., BAMSHAD MICHAEL J. Medical Genetics and Genomics, 2026 (7^a Ed). Editorial Elsevier. ISBN: 978-0443115165. The sixth edition is available in Spanish (Genética médica, 2021, Editorial Elsevier España SL. ISBN 978-84-9113-797-9; eISBN: 978-84-9113-880-8).
- PIERCE B.A. Genetics: A Conceptual Approach. 2024 (7^a Ed.). WH Freeman. ISBN 9781319546700. La fifth edición tiene traducción al castellano (Genética: un enfoque conceptual. 2020. Editorial Médica Panamericana, ISBN 978849110778).
- PRITCHARD D.J. / KORF B.R. Genética Médica. 2015 (3^a 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^a ed, ISBN 0815345895); 2010 (4^a 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:

-ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar/>)

Repository of genetic variants and their clinical interpretation.

-gnomAD (<https://gnomad.broadinstitute.org>)

Repository of exome and genome sequencing data from healthy individuals, providing information on genetic variants, their allele frequencies, and pathogenicity.

-HGMD (Human Gene Mutation Database) (<https://www.hgmd.cf.ac.uk/ac/index.php>)

Database of genetic variants causing Mendelian diseases.

-GeneCards (<http://www.genecards.org/>)

Comprehensive database of annotated and predicted human genes, providing genomic, proteomic, transcriptomic, genetic, and functional information.

-GeneReviews (<http://www.genereviews.org/>)

Expert-authored clinical reviews of inherited disorders, providing up-to-date information on diagnosis, management, and genetic counselling.

-OMIM (Online Mendelian Inheritance in Man) (<https://omim.org/>)

Comprehensive database of human genes and inherited genetic disorders.

-Orphanet (<https://www.orpha.net/>)

European reference portal providing information on rare diseases and orphan drugs.