

**Human Genetics**

Code: 100750  
ECTS Credits: 6

Degree	Type	Year	Semester
2500250 Biology	OT	4	1

**Contact**

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**Use of Languages**

Principal working language: catalan (cat)  
Some groups entirely in English: No  
Some groups entirely in Catalan: No  
Some groups entirely in Spanish: No

**Teachers**

Joan Francesc Barquinero Estruch

**Prerequisites**

To have basic knowledge on genetics.

**Objectives and Contextualisation**

Human Genetics studies the phenomena of heredity and variation both normal and pathological on human species. It is a fundamental and applied subject that integrates all levels of organization, from molecular genetics to evolution genetics.

The main objectives of this course are: understanding the rules and the mechanisms of inheritance, the knowledge of genome variability (normal and pathological) in individuals and human populations and the factors responsible for it, the ability to perform tests for genetic diseases, knowing their treatment and ethical aspects that are derived from treatment, and finally the application of knowledge obtained for development of research projects.

**Competences**

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
- Be able to analyse and synthesise
- Be able to organise and plan.
- Control processes and provide services related to biology.
- Design and carry out biodiagnoses and identify and use bioindicators.
- Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
- Perform genetic analyses.
- Students must be capable of applying their knowledge to their work or vocation in a professional way and they should have building arguments and problem resolution skills within their area of study.

- Students must be capable of collecting and interpreting relevant data (usually within their area of study) in order to make statements that reflect social, scientific or ethical relevant issues.
- Students must be capable of communicating information, ideas, problems and solutions to both specialised and non-specialised audiences.
- Students must develop the necessary learning skills to undertake further training with a high degree of autonomy.
- Students must have and understand knowledge of an area of study built on the basis of general secondary education, and while it relies on some advanced textbooks it also includes some aspects coming from the forefront of its field of study.
- Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
- Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
- Understand heredity mechanisms and the fundamentals of genetic improvement.
- Understand the processes that determine the functioning of living beings in each of their levels of organisation.

## Learning Outcomes

1. Analyse a situation and identify its points for improvement.
2. Analyse the sex- or gender-based inequalities and the gender biases present in one's own area of knowledge.
3. Analyse the sustainability indicators of the academic and professional activities within the area, integrating the social, economic and environmental dimensions.
4. Be able to analyse and synthesise.
5. Be able to organise and plan.
6. Critically analyse the principles, values and procedures that govern the exercise of the profession.
7. Describe heredity patterns and calculate the risk of recurrence of human diseases.
8. Identify the natural and artificial factors that affect human health.
9. Identify the underlying genetic causes of development and of congenital defects in humans.
10. Interact with and advise government institutions operating in the field of social policy and population and public health policy.
11. Interpret human variability as a source of individualisation.
12. Propose new methods or well-founded alternative solutions.
13. Propose projects and actions that incorporate the gender perspective.
14. Propose ways to evaluate projects and actions for improving sustainability.
15. Recognise the anomalies of human chromosomes and assess their consequences.
16. Students must be capable of applying their knowledge to their work or vocation in a professional way and they should have building arguments and problem resolution skills within their area of study.
17. Students must be capable of collecting and interpreting relevant data (usually within their area of study) in order to make statements that reflect social, scientific or ethical relevant issues.
18. Students must be capable of communicating information, ideas, problems and solutions to both specialised and non-specialised audiences.
19. Students must develop the necessary learning skills to undertake further training with a high degree of autonomy.
20. Students must have and understand knowledge of an area of study built on the basis of general secondary education, and while it relies on some advanced textbooks it also includes some aspects coming from the forefront of its field of study.

## Content

Chapter 1. Organization of the human genome

Chapter 2. Chromosomal alterations

Chapter 3. Mutations and polymorphisms

Chapter 4. Epigenetics

Chapter 5. Developmental genetics

Chapter 6. Mapping and identifying genes related to diseases

Chapter 7. Cancer Genetics

Chapter 8. Pharmacogenetics. Nutritional Genomics

Chapter 9. Prenatal Diagnosis. Tests for genetic diseases and genetic counseling.

Chapter 10. Treatment of genetic diseases

Chapter 11. Ethical issues in human genetics

## Methodology

The teaching methodology will benefit from the tools provided by the Virtual Campus of the UAB. To achieve the objectives of the subject, four types of learning activities are proposed: sessions with the all students, seminars with half of the students, autonomous work in groups on an scientific paper and laboratory classes.

Sessions with all students: The students acquire their own knowledge of the subject attending the classes, complementing them with the personal study. These classes are designed as lecture sessions by the teaching staff but also the active participation of students is encouraged to establish discussions or collective reflections. In the classes, digital presentations are used to help the understanding of the contents, which are available on the UAB virtual campus.

Seminars: The knowledge developed in sessions with all students and worked in the personal study is applied to the resolution of practical cases and in the discussion of original research papers published in international journals. Practical cases arise in the form of problems or questions, which are worked on small groups. These type of methodology allow us to reinforce and deepen the topics studied in the sessions with all students.

Autonomous work in small groups on an article: It is proposed to carry out a work in small groups that is prepared outside the classroom and that involves tasks of documentation and group discussion on a topic of human genetics. Tutorials will guide students on how to do this work.

Laboratory practices: It seeks to identify genetic mutations and polymorphisms and relate it to the influence they can have on the generation of diversity and pathological processes. Through these practices the student will acquire skills in the application of instrumental, analytical and molecular techniques.

Annotation: Within the schedule set by the centre or degree programme, 15 minutes of one class will be reserved for students to evaluate their lecturers and their courses or modules through questionnaires.

## Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Laboratory	10	0.4	11, 19, 18, 16, 17, 15, 4, 5
Seminars	10	0.4	6, 3, 2, 1, 7, 8, 9, 11, 14, 12, 20, 19, 18, 16, 17, 15, 4, 5
Sessions with all students	25	1	6, 3, 2, 1, 7, 8, 9, 11, 14, 12, 13, 20, 19, 18, 16, 17, 15, 4, 5
Type: Supervised			
Tutorials	5	0.2	6, 2, 1, 14, 12, 13, 20, 16, 17, 4, 5

Type: Autonomous			
Document research	5	0.2	6, 1, 20, 16, 17, 4, 5
Personal study	45	1.8	1, 7, 8, 9, 11, 20, 19, 16, 17, 15, 4, 5
Problem preparation	15	0.6	1, 7, 8, 9, 11, 20, 18, 16, 17, 15, 4, 5
Report writing	15	0.6	6, 2, 1, 7, 8, 9, 11, 14, 12, 20, 19, 18, 16, 17, 15, 4, 5
Text reading	17	0.68	6, 20, 19, 17, 4, 5

## Assessment

a) Two written tests: each test is 25% of the final mark. The minimum mark to pass the subject will be 5 in each test.

b) Reports from the activities performed in the classes: 20% of the final grade.

c) Work in small groups: 20% of the final mark. In this evaluation we will take into account: the oral presentation (5%), the work (15%) and the adjustment to the limited time. The evaluation of the oral presentation will be individual but the others will be common to all the members of the group.

d) Report related to laboratory classes: 10% of the final mark. Attendance is mandatory to laboratory classes.

To be able to pass the subject, the minimum mark is 5. At the end of the course there will be a remedial test for those students who have failed or not attended any of the two written tests. To be eligible for the retake process, the student should have been previously evaluated in a set of activities equaling at least two thirds of the final score of the course. The student will be graded as "No Avaluable" if the weighthin of all conducted evaluation activities is less than 67% of the final score.

## Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Report related to the laboratory classes	10%	0	0	11, 19, 18, 16, 17, 15, 4
Reports from the activities performed in the classes	20%	0	0	6, 3, 2, 1, 7, 8, 9, 10, 11, 14, 12, 13, 20, 19, 18, 16, 17, 15, 4, 5
Work in group	20%	0	0	7, 9, 11, 15, 4, 5
Written test I	25%	1.5	0.06	7, 8, 9, 11, 15, 4, 5
Written test II	25%	1.5	0.06	7, 8, 9, 11, 15, 4, 5

## Bibliography

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- <http://www.ncbi.nlm.nih.gov/omim>
- <http://ghr.nlm.nih.gov>
- <http://www.genome.gov>

## Software

No specific software will be used.