# Human Genetics

Code: 100750
ECTS Credits: 6

**2024/2025**

| Degree | Type | Year |
|---|---|---|
| 2500004 Biology | OT | 4 |

## Contact

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## Teachers

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## Teaching groups languages

You can view this information at the end of this document.

## 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.
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## 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

## Activities and Methodology

| 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, 8, 9, 11, 14, 12, 20, 19, 18, 16, 17, 15, 4, 5 |
| Sessions with all students | 25 | 1 | 6, 3, 2, 1, 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 |

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

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, using Information and Communications Technologies and Learning and Knowing Technologies. 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 cooperative 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 sessions: 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. The attendance to the laboratory sessions is compulsory.

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.

## Assessment

## Continous 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 |

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.

Unique assessment
Students enrolled in the single assessment will have to take the two written tests and hand in the assignments from works performed in the classes on a single date, only for those that are not in group (under the same conditions as students with continuous assessment). The works performed in the classes that are in group and the work in small groups will be assessed on the same day that students with continuous assessment. Laboratory classes are of mandatory attendance and will be assessed on the same day that students with continuous assessment: the last day of laboratory classes. The single assessment test will coincide with the same date fixed in the calendar for the last continuous assessment test and the same recovery system will be applied as for the continuous assessment.

## 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.

## Language list

| Name | Group | Language | Semester | Turn |
|---|---|---|---|---|
| (PAUL) Classroom practices | 141 | Catalan | first semester | morning-mixed |
| (PLAB) Practical laboratories | 141 | Catalan | first semester | afternoon |
| (PLAB) Practical laboratories | 142 | Catalan | first semester | afternoon |
| (TE) Theory | 14 | Catalan | first semester | morning-mixed |