

Human Genetics

Code: 100750
ECTS Credits: 6

Degree	Type	Year	Semester
2500250 Biology	OT	4	0

Contact

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Use of languages

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

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.

Skills

- 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.
- Develop critical thinking and reasoning and communicate ideas effectively, both in the mother tongue and in other languages.
- Develop independent learning strategies.
- Perform genetic analyses.
- Respect diversity in ideas, people and situations
- 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. Be able to analyse and synthesise.

2. Be able to organise and plan.
3. Describe heredity patterns and calculate the risk of recurrence of human diseases.
4. Develop critical thinking and reasoning and communicate ideas effectively, both in the mother tongue and in other languages.
5. Develop independent learning strategies.
6. Identify the natural and artificial factors that affect human health.
7. Identify the underlying genetic causes of development and of congenital defects in humans.
8. Interact with and advise government institutions operating in the field of social policy and population and public health policy.
9. Interpret human variability as a source of individualisation.
10. Recognise the anomalies of human chromosomes and assess their consequences.
11. Respect diversity in ideas, people and situations.

Content

Chapter 1. Organization of the human genome

Chapter 2. Chromosomal alterations

Chapter 3. Mutations and polymorphisms .

Chapter 4. Mapping and identifying genes related to diseases

Chapter 5. Epigenetics

Chapter 6. Developmental genetics

Chapter 7. Cancer Genetics

Chapter 8. Pharmacogenetics

Chapter 9. Nutritional Genomics

Chapter 10. Prenatal Diagnosis

Chapter 11. Forensic Genetics

Chapter 12. Tests for genetic diseases and genetic counseling

Chapter 13. Treatment of genetic diseases

Chapter 14. 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, three types of learning activities are proposed: theoretical sessions, seminars and autonomous work in groups on a topic.

Theoretical sessions: The students acquire their own knowledge of the subject attending the classes of theory, 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 theory classes 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 theoretical sessions.

Autonomous work in groups on a topic: It is proposed the realization of 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.

Activities

Title	Hours	ECTS	Learning outcomes
Type: Directed			
Seminars	15	0.6	3, 5, 4, 7, 9, 10, 11, 1, 2
Theoretical sessions	30	1.2	3, 4, 7, 9, 10
Type: Supervised			
Tutorials	5	0.2	5, 4, 11, 1, 2
Type: Autonomous			
Document research	5	0.2	5, 4, 1, 2
Personal study	45	1.8	3, 7, 9, 10, 1, 2
Problem preparation	15	0.6	3, 5, 7, 9, 10, 11, 1, 2
Report writing	15	0.6	3, 5, 4, 7, 9, 10, 11, 1, 2
Text reading	17	0.68	5, 4, 1, 2

Evaluation

a) Two written tests: each test is 30% of the final mark. The minimum mark to pass the subject will be 5 in each test.

b) Resolution of problems and comments on scientific articles (in the seminar sessions): 20% of the final grade.

c) Working in 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.

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.**

Evaluation activities

Title	Weighting	Hours	ECTS	Learning outcomes
Resolution of problems and comments on scientific articles	20%	0	0	3, 5, 4, 7, 8, 9, 10, 11, 1, 2
Working in groups	20%	0	0	3, 5, 4, 7, 9, 10, 11, 1, 2

Written test I	30%	1.5	0.06	3, 5, 6, 7, 9, 10, 1, 2
Written test II	30%	1.5	0.06	3, 5, 6, 7, 9, 10, 1, 2

Bibliography

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