Universitat Autònoma de Barcelona

2023/2024

Human Genetics

Code: 101971
ECTS Credits: 6

Degree	Type	Year	Semester
2500890 Genetics	OB	3	1

Contact

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Teaching groups languages

You can check it through this link. To consult the language you will need to enter the CODE of the subject. Please note that this information is provisional until 30 November 2023.

Prerequisites

To have some 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.
- Appreciate the importance of quality and a job well done.
- Be able to analyse and synthesise.
- Be able to communicate effectively, orally and in writing.
- Define mutation and its types, and determine the levels of genic, chromosomal and genomic damage in the hereditary material of any species, both spontaneous and induced, and evaluate the consequences.
- Describe and interpret the principles of the transmission of genetic information across generations.

- Describe the genetic bases of the development and control of genic expression.
- Design and interpret studies associating genetic polymorphisms and phenotypical characters to identify genetic variants that affect the phenotype, including those associated to pathologies and those that confer susceptibility to human illnesses or those of other species of interest.
- Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
- Measure and interpret the genetic variation in and between populations from a clinical, conservational and evolutionary perspective, and from that of the genetic improvement of animals and plants.
- Perform genetic diagnoses and assessments and consider the ethical and legal dilemmas.
- Produce, direct, execute and assess projects where knowledge of genetics or genomics is necessary.
- 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.
- Take the initiative and demonstrate an entrepreneurial spirit.
- Use and interpret data sources on the genomes and macromolecules of any species and understand the basics of bioinformatics analysis to establish the corresponding relations between structure, function and evolution.

Learning Outcomes

1. Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
2. Appreciate the importance of quality and a job well done.
3. Be able to analyse and synthesise.
4. Be able to communicate effectively, orally and in writing.
5. Describe the genetic basis of human development.
6. Describe the role of genetic variation in the human species in the diagnoses, prevention and treatment of illnesses.
7. Determine the genetic basis and calculate the risk of recurrence of human illnesses.
8. Enumerate and describe the different techniques for analysing DNA polymorphisms that can be applied to studies of genetic variation associated to human pathologies.
9. Interpret scientific publications, and solve problems and example cases in the fields of human and cancer genetics.
10. Interpret the results obtained using techniques for the analysis of DNA polymorphisms to identify and evaluate factors of susceptibility and propensity to suffer illnesses.
11. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
12. Propose genetics or genomics projects that are applicable to the field of human health.
13. Recognise genic, chromosomal and genomic anomalies in humans and evaluate the clinical consequences.
14. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
15. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
16. Take the initiative and demonstrate an entrepreneurial spirit.
17. Use data sources on the human genome and interpret them.

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. Forensic Genetics

Chapter 11. Prenatal Diagnosis

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: 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.

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			
Seminars	15	0.6	16, 4, 3, 2

Sessions with all students	30	1.2	16, 4
Type: Supervised			
Tutorials	5	0.2	16, 3, 2
Type: Autonomous			
Document research	5	0.2	16, 3, 2
Personal study	45	1.8	3, 2
Problem preparation	15	0.6	3
Report writing	15	0.6	16, 4, 3, 2
Text reading	17	0.68	3, 2

Assessment

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) Handling works performed in the classes: 20% of the final grade. There are evaluable works in some classes.

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.

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 the students with continuous assessment will be assessed. 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.

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Handling works performed in the classes	20%	0	0	1, 15, 14, 5, 7, 8, 10, 9, 11, 16, 12, 13, 4, 3, 17, 2
Working in groups	20%	0	0	1, 15, 14, 6, 5, 7, 8, 10, 9, 11, 16, 12, 13, 4, 3, 17, 2
Written test I	30%	1.5	0.06	6, 5, 7, 8, 10, 13, 3, 17, 2

Written test II	30%	1.5	0.06	6, 5, 7, 8, 10, 13, 3, 17, 2

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