Human Genetics

Code: 101887
ECTS Credits: 6

2024/2025

Degree	Type	Year
2501230 Biomedical Sciences	OB	2

Contact

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Teachers

Joan Francesc Barquinero Estruch

Teaching groups languages

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

Prerequisites

Those needed to follow the degree, and have taken the subject of genetics during the first year. It is not necessary to have passed the genetics course.

Objectives and Contextualisation

Human Genetics studies the phenomena of inheritance for both normal and pathological variation in humans. It is a fundamental and applied subject in biomedical sciences that integrates all levels of organization, from the molecular to the evolutionary.

The main objectives of this subject are:

1. Understanding the bases and mechanisms of inheritance.

2. Ability to perform genetic analyses of different characters.

3. Ability to design and obtain information from experiments in genetics, as well as to interpret the results obtained.

4. To develop of a historical vision that allows summarizing the most important milestones of Human Genetics and assess the contributions to current biology and medicine.

Competences

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
- Display knowledge of the bases and elements applicable to the development and validation of diagnostic and therapeutic techniques.
- Display knowledge of the basic life processes on several levels of organisation: molecular, cellular, tissues, organs, individual and populations.
- Display knowledge of the concepts and language of biomedical sciences in order to follow biomedical literature correctly.
- Display theoretical and practical knowledge of the major molecular and cellular bases of human and animal pathologies.
- Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
- Read and critically analyse original and review papers on biomedical issues and assess and choose the appropriate methodological descriptions for biomedical laboratory research work.
- 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.
- Work as part of a group with members of other professions, understanding their viewpoint and establishing a constructive collaboration.

Learning Outcomes

1. Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
2. Contrast the techniques and methods that allow genetic diagnosis.
3. Correctly use the terminology of genetics and its text and reference books
4. Describe and understand the genetic bases of sex determination and differentiation in humans.
5. Describe the genetic bases of cancer.
6. Describe the organisation, evolution, inter-individual variation and expression of the human genome.
7. Design methodologies for the experimental study of genetic diseases.
8. Identify chromosome variants and anomalies, understand the mechanisms that originate them and determine the risk of their transmission to descendents.
9. Identify the genetic bases of human development.
10. Identify the genetic bases of the principal diseases with a genetic base or component.
11. Interpret genetically the diagnosis, prognosis, prevention and therapy for the most frequent genetic pathologies in the human population.
12. Interpret scientific publications and solve problems and typical cases in the area of cytogenetics.
13. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
14. Provide pre-conceptional genetic counselling, taking ethical and legal factors into account.
15. Recognise and identify the distribution of genetics-based diseases in a particular population, taking the origin into account.
16. Recognise the anomalies of human chromosomes and assess their consequences.

17. Relate genetic dysfunction to the pathological phenotype.
18. 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.
19. 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.
20. Students must be capable of communicating information, ideas, problems and solutions to both specialised and non-specialised audiences.
21. Students must develop the necessary learning skills to undertake further training with a high degree of autonomy.
22. 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.
23. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
24. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
25. Understand scientific texts on genetics and development, and write review papers on them.
26. Work as part of a group with members of other professions, understanding their viewpoint and establishing a constructive collaboration.

Content

Topic 1. The human genome

Topic 2. Developmental Genetics

Topic 3. Mutations and polymorphisms

Topic 4. Chromosomal alterations

Topic 5. Gene mapping

Topic 6. Pharmacogenetics and nutritional genomics

Topic 7. Cancer genetics

Topic 8. Genetic counseling and ethics

Topic 9. Treatment of genetic diseases: gene therapy

In case of inconsistency between the different teaching guides, the one that prevails is the teaching guide in Catalan

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
In-person classes	37	1.48	2, 4, 6, 5, 7, 10, 9, 8, 11, 12, 14, 15, 16, 17, 3
Seminars	9	0.36	25, 12, 26
Type: Supervised			

Individual tutoring	4	0.16	
Type: Autonomous			
Self-study	89.5	3.58	25, 2, 4, 6, 5, 10, 9, 8, 11, 14, 15, 16, 17, 3
Team work	8	0.32	25, 2, 4, 6, 5, 7, 10, 9, 8, 11, 12, 14, 15, 16, 17, 26, 3

Contents of the subject are oriented to understand: the organization of the human genome; the organization, distribution and function of the RNA genes and genes encoding polypeptides; Tandem and scattered repeat non-coding DNA; Genetics of development; genetic control of embryonic development; congenital defects, sex determination and differentiation; genetic imprinting; inactivation of the X chromosome; Genetic of populations, mutation and genetic polymorphisms in human populations; Human cytogenetics, methods of study and main chromosomal alterations; Genetics and cancer, oncogenes, tumor suppressor genes, and genome stability; Prenatal diagnosis: indications and study techniques, prenatal screenings. Genetic tests in individuals and populations. General diagnostic strategies for genetic diseases. Mutation detection methods. Application of the genetic ligament to diagnosis: indirect diagnosis.

In-person classes: the student acquires the scientific knowledge of the subject by attending theory classes, which will complement the personal study of the topics discussed. Classes are considered unidirectional as a transmission from teacher to student. Although at certain moments a debate or collective reflection is possible.

Classes of problems and seminars: The knowledge developed in the theory classes and worked on in the personal study are applied to the resolution of practical cases and oral presentations in small groups. These types of methodology allow to deepen some of the topics studied in class.

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
Individual tasks during the academic course	5%	0	0	2, 4, 6, 5, 10, 9, 8, 11, 12, 21, 20, 19, 14, 15, 16, 17, 3
Solvig practical problems, answer short questions, comments on scientific papers	20%	0.5	0.02	1, 24, 12, 22, 21, 18, 19, 26
Work team presentation	15%	0	0	1, 24, 23, 12, 13, 22, 20, 18, 19, 26, 3
mid-term exams, and final exam	60%	2	0.08	1, 23, 25, 2, 4, 6, 5, 7, 10, 9, 8, 11, 13, 14, 15, 16, 17, 3

a) Two partial exams, test type and/or with short questions. Each partial will include approximately 50% of the topics.

In the partial exams of human genetics, the minimum grade to make an average will be a 5. At the end of the year there will be a recovery test, for those students who have not passed any of the partial exams, or who have not submitted to any of the two written tests.

The average grade of these exams represents 60% of the final grade. Students who have passed the two partial exams and want to raise their grade, can take an exam on the same day of the recovery, which will consist of developing two or three subjects

b) Problem solving exam: 20% of the final mark.

c) Work prepared in groups of four students: 15% of the final grade. This evaluation will take into account: the contents (10%) the oral presentation (5%). The evaluation will be individual.

d) The resolution of small tasks during the course: 5% of the final mark

It will be considered that a student will obtain the grade of Non-Evaluable if, not taking the single assessment, the number of assessment activities carried out has a weight of less than 50% of the final grade.

Unique assessment:

The single assessment consists of a test that includes a synthesis test in which the contents of the entire theory program, classroom practices and seminars will be evaluated (this part has a weight of 75% of the grade), and a test in which the summary of a scientific article will be defended using an oral presentation (this part has a weight of 25% of the grade). The synthesis test will consist of test-type questions or short questions.

In case of inconsistency between the different teachers, the teaching guide in Catalan prevails.

Bibliography

Lynn B. Jorde, John C. Carey, Michael J. Bamshad (2016). Medical Genetics. Fihth Edition. Ed Mosby. Elsevier Science. ISBN 978-0-323-39196-2

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Strachan T and Godship J Chinnay P (2015). Genetics and genomics in medicine. 5ª Ed, Garland Science London UK

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Solari AJ (2011). Genética Humana. Fundamentos y aplicaciones en medicina. 4ª edición Editorial Médica Panamericana. Buenos Aires. ISBN: 9789500602693

Tobias ES, Connor M, Ferguson-Smith M (2011). Essential MEDICAL GENETICS.6th Ed.Wiley-Blacwell. ISBN: 978-1-405-16974-5

Speicher MR Antonarakis SE Motulsky AG (2010). Vogel and Motulsky's Human Genetics: Problems and Approaches. 4th ed Springer-Verlag. Berlin.

•http://www.ncbi.nlm.nih.gov/omim/ Enfermedades con base hereditaria

•http://bioinformatics.weizmann.ac.il/cards/

•http://www.ncbi.nlm.nih.gov/PubMed/

•http://www. Genome.gov

www.gdb.org Datos procedentes del proyecto del genoma humano

geneReviews.org Información completa de enfermedades genéticas

orphanet Información de enfermedades genéticas

Software

There is no specific programmes.

Language list

Name	Group	Language	Semester	Turn
(SEM) Seminars	521	Catalan/Spanish	first semester	morning-mixed
(SEM) Seminars	522	Catalan/Spanish	first semester	morning-mixed
(TE) Theory	52	Catalan	first semester	afternoon