Universitat Autònoma de Barcelona

2019/2020

Human Genetics

Code: 101887
ECTS Credits: 6

Degree	Type	Year	Semester
2501230 Biomedical Sciences	OB	2	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: Yes

Teachers

Joan Francesc Barquinero Estruch

Prerequisites

Those needed to follow the degree, and have taken the subject of genetics during the first year

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

- Contribute to public discussions on cultural matters.
- Develop critical thinking and reasoning and communicate ideas effectively, both in the mother tongue and in other languages.
- Develop independent learning habits and motivation to continue training at postgraduate level.
- Develop independent learning strategies.

- Develop scientific knowledge, critical reasoning and creativity.
- 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.
- Generate innovative and competitive proposals for research and professional activities.
- Identify and understand the advances and challenges of research.
- Plan and implement laboratory analysis experiments and procedures belonging to the biomedical field.
- Read and critically analyse original and review papers on biomedical issues and assess and choose the appropriate methodological descriptions for biomedical laboratory research work.
- Respect diversity in ideas, people and situations.
- Show respect for the ethical and legal aspects of research and professional activities.
- Work as part of a group with members of other professions, understanding their viewpoint and establishing a constructive collaboration.

Learning Outcomes

1. Contrast the techniques and methods that allow genetic diagnosis.
2. Contribute to public discussions on cultural matters.
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. Develop critical thinking and reasoning and communicate ideas effectively, both in the mother tongue and in other languages.
9. Develop independent learning habits and motivation to continue training at postgraduate level.
10. Develop independent learning strategies.
11. Develop scientific knowledge, critical reasoning and creativity.
12. Generate innovative and competitive proposals for research and professional activities.
13. Identify and understand the advances and challenges of research.
14. Identify chromosome variants and anomalies, understand the mechanisms that originate them and determine the risk of their transmission to descendents.
15. Identify the genetic bases of human development.
16. Identify the genetic bases of the principal diseases with a genetic base or component.
17. Interpret genetically the diagnosis, prognosis, prevention and therapy for the most frequent genetic pathologies in the human population.
18. Interpret scientific publications and solve problems and typical cases in the area of cytogenetics.
19. Provide pre-conceptional genetic counselling, taking ethical and legal factors into account.
20. Recognise and identify the distribution of genetics-based diseases in a particular population, taking the origin into account.
21. Recognise the anomalies of human chromosomes and assess their consequences.
22. Relate genetic dysfunction to the pathological phenotype.
23. Respect diversity in ideas, people and situations.
24. Show respect for the ethical and legal aspects of research and professional activities.
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. Pharmacogenetics and nutritional genomics

Topic 6. Mapping and identification of genes

Topic 7. Cancer genetics

Topic 8. Genetic counseling and prenatal diagnosis

Topic 9. Genetic analyzes in individuals and in human populations

Topic 10. Treatment of genetic diseases: gene therapy

Methodology

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.

Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
In-person classes	37	1.48	24, 1, 4, 6, 5, 11, 10, 9, 7, 13, 16, 15, 14, 17, 18, 19, 20, 21, 22, 23, 3
Seminars	9	0.36	25, 11, 10, 9, 8, 12, 13, 18, 23, 26
Type: Supervised			
Individual tutoring	4	0.16	
Type: Autonomous			
Self-study	89.5	3.58	25, 1, 4, 6, 5, 11, 10, 9, 13, 16, 15, 14, 17, 19, 20, 21, 22, 3
Team work	8	0.32	24, 25, 2, 1, 4, 6, 5, 11, 10, 9, 8, 7, 12, 13, 16, 15, 14, 17, 18, 19, 20, 21, 22, 23,

Assessment

a) Two mid-term examinations. These are multiple choice examinations in which short questions can be included. These exams are eliminatory. Each mid-term exam will include approximately 50% of the topics.

In the mid-term exams the minimum mark to be considered for the mean is 5. At the end of the course there is a recovery exam for those students who have not passed some of the partial exams (with marks less than 5), or have not examined to any of the two multiple choice tests.

The average mark of these exams represents 60% of the note mark.

b) Examining practical problems: 20% of the final grade.

c) Work prepared in groups of four students: 20% of the final grade. This evaluation will consider: the contents (15%) the oral presentation (5%. The evaluation is individual.

It will be considered that a student will obtain the qualification of "No Avaluable" if the number of assessment activities carried out has a weight inferior to 50% of the final grade.

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Solvig practical problems, answer short questions, comments on scientific papers	20%	2.5	0.1	9, 18
Work team presentation	20%	0	0	2, 11, 10, 9, 13, 18, 26, 3
mid-term exams, and final exam	60%	0	0	24, 25, 1, 4, 6, 5, 11, 8, 7, 12, 16, 15, 14, 17, 19, 20, 21, 22, 23, 3

Bibliography

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Strachan T and Read A. (2018). Human Molecular Genetics. CRC Press. Taylor and Francis. ISBN 9780815345893

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Oliva R Oriola F Clària J (2013).Genètica Mèdica. Publicacions i Edicions Universitat de Barcelona. ISBN: 978-84-475-3688-7

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

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•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