Medical Genetics

Code: 101970
ECTS Credits: 6

2025/2026

Degree	Type	Year
Genetics	OB	3

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Teachers

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Teaching groups languages

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

Prerequisites

It would be appropriate to have passed and achieved the competences corresponding to the subjects: Cell Biology, Human Genetics, Molecular Cell Biology, Developmental Biology and Teratogeny.

Objectives and Contextualisation

The main objectives are:

Know the genetic basis of the main diseases with a base or genetic component.
Relate the genetic dysfunction with the pathological phenotype.
Perform the genetic interpretation of the diagnosis, prognosis, prevention and therapy of the most frequent genetic pathologies in the human population.
Understand the distribution of genetic-based diseases in a population taking into account their origin.
Analyze genetically the probands-family relationship that facilitates the offert of a genetic counseling.

Competences

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
- Apply knowledge of theory to practice.
- Appreciate the importance of quality and a job well done.
- Assume ethical commitment
- Be able to analyse and synthesise.
- Be sensitive to environmental, health and social matters.
- Describe epigenetic mechanisms.
- Describe the genetic bases of the development and control of genic expression.
- Describe the organisation, evolution, inter-individual variation and expression of the human genome.
- 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.
- Develop analysis, synthesis and communication strategies to transmit the different aspects of genetics in educational settings.
- 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.
- Perceive the strategic, industrial and economic importance of genetics and genomics to life sciences, health and society.
- Perform genetic diagnoses and assessments and consider the ethical and legal dilemmas.
- 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.

Learning Outcomes

1. Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
2. Apply knowledge of theory to practice.
3. Appreciate the importance of quality and a job well done.
4. Assume ethical commitment
5. Be able to analyse and synthesise.
6. Be sensitive to environmental, health and social matters.
7. Describe the clinical consequences derived from epigenetic control mechanisms.
8. Describe the genetic and environmental causes of congenital defects.
9. Describe the role of genetic variation in the human species in the diagnoses, prevention and treatment of illnesses.
10. Describe the structure and variation of the human genome from a functional, clinical and evolutionary perspective.
11. Enumerate and describe the different techniques for analysing DNA polymorphisms that can be applied to studies of genetic variation associated to human pathologies.
12. Evaluate and interpret genetic variation in human populations and from a clinical and evolutionary perspective.
13. Explain how knowledge of human genetic variation is applied to personalised medicine, pharmacogenomics and nutrigenomics.
14. Interpret scientific publications, and solve problems and example cases in the fields of human and cancer genetics.
15. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
16. Prepare communication proposals in educational settings regarding the importance of the transfer of genetic progress to clinical practice.
17. Recognise the strategic importance of genetic progress in the field of human health, especially applications of the genomic to personalised medicine, pharmacogenomics and nutrigenomics.

18. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
19. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.

Content

1. General principles: Self-learning activity
1.1. Basic introduction to genetic diseases
1.2. Predisposition or genetic susceptibility concept
1.3. Genetic counseling

2. Chromosome aberrations
2.1. Autosomal trisomies
2.2. Alterations of sex chromosomes
2.3. Chromosome microdeletions and microduplications

3. Neuromuscular diseases
3.1. Muscular dystrophies: definition and classification
3.2. Duchenne and Becker muscular dystrophies
3.3. Other muscular dystrophies
3.4. Myotonic dystrophy
3.5. Spinal muscular atrophy

4. Mental and behavioural disorders
4.1. Intellectual Disability
4.2. Fragile X syndrome and associated diseases
4.3. Rett syndrome
4.4. Introductio to the genetics of autism spectrum disorder (ASD)
4.5. Introduction to genetics of Schizophrenia

5. Central nervous system diseases
5.1. Huntington's disease
5.2. Alzheimer's disease
5.3. Parkinson's
5.4. Primary tumors of the central nervous system

6. Connective tissue diseases
6.1. Marfan syndrome
6.2. Osteogenesis imperfecta
6.3. Sarcoma

7. Craniofacial and skeletal diseases
7.1. Craniosynostosis
7.2. Achondropasia

8. Dermatological diseases
8.1. Albinism
8.2. Skin cancer

9. Ophthalmological diseases and deafness
9.1. Color vision deficiency
9.2. Deafness

10. Cardiovascular diseases
10.1. Hypertension

11. Gastrointestinal diseases
11.1. Celiac and inflammatory diseases
11.2. Colorectal cancer

12. Respiratory diseases
12.1. Cystic fibrosis

13. Metabolic diseases
13.1. Alterations in glucose metabolism: lactose intolerance
13.2. Alterations in lipid metabolism: Hypercholesterolemia
13.3. Alterations in amino acid metabolism: Phenylketonuria

14.Hematological diseases
14.1. Hemoglobinopathies
14.2. Haemophilia
14.3. Leukemia and Lymphoma

15. Urogenital diseases
15.1. Polycystic kidney
15.2. Kidney cancer
15.3. Bladder cancer

16. Endocrinological diseases
16.1. Breast cancer
16.2. Diabetes mellitus

17. Imprinting-related diseases
17.1. Beckwith-Wiedemann and Silver-Russell

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Lectures	34	1.36	4
Problem based learning (PBL)	12	0.48	2, 4, 6, 5, 3
Type: Supervised			
Face-to-face and virtual tutorials	6	0.24	2, 4
Work production	18	0.72	2, 4, 6, 5, 3
Type: Autonomous			
Documentation search	17	0.68	
Evaluation	6	0.24	
Reading of texts	19	0.76	5
Study	38	1.52	5

Lectures: Systematic exhibition of the subject's programme, giving relevance to the most important concepts. The student acquires the basic scientific knowledge of the subject in theory classes, which will complement the personal study of the exposed themes.

Problem based learning (PBL): Students will work in small groups, under the teacher supervision, on specific problems during 3 sessions of 2 hours each one for each case, and a total of 2 cases. At the end of each case, the work will be exposed to the rest of the classmates.

In general, the platform for communication and material transfer used will be Moodle.

"Teachers should allocate approximately 15 minutes of some class to allow their students to answer the surveys of evaluation of teaching performance and evaluation of the subject or module"

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
Exams	75%	0	0	1, 19, 18, 12, 9, 8, 7, 10, 13, 15, 17
Problem based learning (PBL)	25%	0	0	1, 19, 18, 2, 4, 6, 11, 14, 15, 16, 5, 3

Thissubject does not include single assessment.

The competences of this subject will be evaluated through: exams, group work and public presentations.

The evaluation system is organized in two modules, each of which will have a specific weight assigned in the final qualification:

- Problem Based Learning Module (ABP) (25%). The aspects that will be taken into account for the qualification will be: the interest and the quality of the work demonstrated throughout the development of the case for each one of the students and the group, and the final presentation. In the case of repeating students, if the qualification of this learning evidence in previous courses was equal to or greater than 5, they can renounce to repeat the activity and this qualification will be used to calculate the global grade of the subject. Due to the characteristics of the activity, this learning evidence is not recoverable.
- Written test module (75%). There will be two exams, each one corresponding to a half of the subject taught in lecture sessions. Exams will be test type with four multiple-choice options.

Evaluation activities	Recoverable	Value
1st part	Si	37,5%
2nd part	Si	37,5%

PBL case 1	No	12,5%
PBL case 2	No	12,5
Total		100%

Requirements to calculate the global qualification

The weighted average of the scores will be applied only if a grade equal to or higher than 4 is obtained in each of the partial exams.

In order to calculate an average between the partial exams and the PBLs (Problem-Based Learning activities), and consequently to pass the course, a minimum average grade of 5 in the partial exams must be achieved.

The grade "Not Assessable" will be given if the number of assessment activities completed is less than 50% of those scheduled for the course.

Recovery exam / retake process

To participate in the recovery (resit) exam, students must have previously been assessed in a set of activities whose weight is equivalent to at least two-thirds of the total grade for the course. Therefore, students will receive the grade "Not Assessable" if the completed assessment activities account for less than 67% of the final grade.

Partial exams in which the exam score was below 4 can be retaken.

The format of the resit exams may include multiple-choice questions and/or short-answer questions to be developed by the student.

Students who wish to improve the grade of one or both partial exams may do so in the resit exam, but must first waive the previously obtained grade in the corresponding partial exam.

Copy and plagiarism

Copying or plagiarism, whether in assignments or in exams, constitutes an offense that may result in failing the course.

Bibliography

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- Lee, R.J., Abramson, J.S I Goldson, R.A. Case studies in cancer. Ed. W. W. Norton & Company. 2019
- Read A. i Donnai D. New Clinical Genetics. Editorial Scion Publishing Ltd, 2011
- Rimon D.L. Emery and Rimoin's principles and practice of medical genetics, 5a ed. Editorial Churchill Livingstone. Vol 2-3, 2006
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- Scriver Ch.R., et al. The Metabolic & molecular bases of inherited disease, 8th ed. Editorial MacGraw-Hill, NewYork, 2001
- Stracher T., et al. Genetics and Genomics in Medicine, 1st ed. Garland Science, 2014
- Weinberg, R.A. The Biology of Cancer. Garland Science, 2a ed. (2014)
- eBooks:
- - The Biology of Cancer
  - Molecular and Cell Biology of Cancer
  - Molecular Genetics & Genomic Medicine
  - Genomic and Personalized Medicine
  - Thompson y Thompson. Genética en medicina (8a. ed.)
  - Human Chromosomes: an Illustrated Introduction to Human Cytogenetics.
  - Cytogenetic and genome research
  - Haemoglobinopathy Diagnosis
  - Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling
  - Oxford Desk Reference: Clinical Genetics and Genomics
  - Harper's Practical Genetic Counselling, Eighth Edition
  - Essentials of genomic and personalized medicine [Recurs electrònic]
  - Genética Médica

Software

To carry out the theoretical classes, programs from the Miscrosoft package will be used, essentially PowerPoint, Adobe pdf, and if videoconferences are necessary, Teams will be used.

Groups and Languages

Please note that this information is provisional until 30 November 2025. You can check it through this link. To consult the language you will need to enter the CODE of the subject.

Name	Group	Language	Semester	Turn
(SEM) Seminars	631	Catalan	second semester	afternoon
(SEM) Seminars	632	Catalan	second semester	morning-mixed
(SEM) Seminars	633	Catalan	second semester	morning-mixed
(TE) Theory	63	Catalan	second semester	morning-mixed