

Degree	Type	Year
Biomedical Sciences	OB	3

Contact

Name: Jordi Camps Polo

Email: jordi.camps@uab.cat

Teachers

Jordi Ribas Maynou

Maria Angels Rigola Tor

Maria Oliver Bonet

Jordi Camps Polo

Alejandro Gella Concustell

Mariona Terradas III

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.
- Display knowledge of the bases and elements applicable to the development and validation of diagnostic and therapeutic techniques.
- 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. Design methodologies for the experimental study of genetic diseases.
5. Identify the genetic bases of the principal diseases with a genetic base or component.
6. Interpret scientific publications and solve problems and typical cases in the area of cytogenetics.
7. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
8. Relate genetic dysfunction to the pathological phenotype.
9. 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.
10. 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.
11. Students must be capable of communicating information, ideas, problems and solutions to both specialised and non-specialised audiences.
12. Students must develop the necessary learning skills to undertake further training with a high degree of autonomy.
13. 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.
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. Understand scientific texts on genetics and development, and write review papers on them.
17. Work as part of a group with members of other professions, understanding their viewpoint and establishing a constructive collaboration.

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. Autism spectrum disorder (ASD)
 - 4.5. 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 Achondroplasia
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 aminoacid 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	16, 2, 5, 8, 3
Problem based learning (PBL)	12	0.48	16, 2, 4, 5, 6, 8, 17, 3
Type: Supervised			
Face-to-face and virtual tutorials	6	0.24	5, 8
Work production	18	0.72	16, 2, 4, 5, 6, 8, 17, 3
Type: Autonomous			
Documentation search	17	0.68	16, 2, 6
Evaluation	6	0.24	1, 15, 14, 2, 5, 6, 7, 13, 12, 11, 9, 10, 8, 3
Reading of texts	19	0.76	16, 2, 5, 6, 8, 3
Study	38	1.52	16, 2, 4, 5, 6, 8, 17, 3

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

Continuous Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Exams	75%	0	0	16, 2, 4, 5, 6, 8, 3
Problem based learning (PBL)	25%	0	0	1, 15, 14, 16, 2, 4, 5, 6, 7, 13, 12, 11, 9, 10, 8, 17, 3

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.

EXAMINATION-BASED (single) ASSESSMENT

The two problem-based learning sessions (APBs) are compulsory for all students, including those enrolling to the single assessment option. The evaluation and weighting of these sessions on the final grade will be the same as those of the continuous evaluation (ABP case 1: 12.5%; APB case 2:12.5%).

Single assessment consists of a single examination (with multiple-choice questions) on the contents of the entire theory programme.

The mark obtained in this test is 75% of the final grade of the subject.

The single assessment examination will coincide with the same date established in the calendar for the last continuous assessment test and the same recovery system will be applied as for continuous assessment.

To pass the subject, a minimum score of 4 points out of 10 must be obtained in the multiple-choice test in order to be able to average with the marks obtained in the APB sessions.

Students who wish to improve the qualification of one or both partial exams may do it in the recovery exam, previously renouncing to the qualification obtained 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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- 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

This subject does not require any specific software

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	531	Catalan	second semester	afternoon

(SEM) Seminars	532	Catalan	second semester	afternoon
(SEM) Seminars	533	Catalan	second semester	afternoon
(TE) Theory	53	Catalan	second semester	morning-mixed