Universitat Autònoma de Barcelona

2020/2021

Medical Genetics

Code: 101886
ECTS Credits: 6

Degree	Type	Year	Semester
2501230 Biomedical Sciences	OB	3	2

The proposed teaching and assessment methodology that appear in the guide may be subject to changes as a result of the restrictions to face-to-face class attendance imposed by the health authorities.

Contact

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Use of Languages

Principal working language: catalan (cat)
Some groups entirely in English: No
Some groups entirely in Catalan: Yes
Some groups entirely in Spanish: No

Teachers

Vicenç Català Cahís
Immaculada Ponsa Arjona

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
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
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. Skeletal and connective tissue diseases
6.1. Osteogenesis imperfecta
6.2. Achondroplasia
6.3. Marfan syndrome
6.4. Sarcoma

7. Craniofacial diseases
7.1. Craniosynostosis

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 disease
11.2. Colon cancer

12. Respiratory diseases
12.1. Lung cancer

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
15.4. Prostate cancer

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

Note: ***Unless the requirements enforced by the health authorities demand a prioritization or reduction of these contents***

Methodology

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.

Note: The proposed teaching methodology may experience some modifications depending on the restrictions to face-to-face activities enforced by health authorities.

Activities

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

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 based on qualifications equal to or greater than 4 in each of the partial exams.

In order to pass the subject, it will be necessary to obtain a global qualification equal to or greater than 5.

Recovery exam / retake process
To be eligible for the retake process, the student should have been previously evaluated in a set of activities equalling at least two thirds of the final score of the course. Thus, the student will be graded as "No Avaluable" if the weighting of all conducted evaluation activities is less than 67% of the final score.

The partial exams can be recovered when the qualification of the exam has been less than 4. The student can do the recovery exam only of the partial suspended.

If the student has to recover both partial exams, the score obtained corresponds to 75% of the qualification of the subject, disappearing the requirement of having to obtain a score greater than or equal to 4 of each of the partial exams. This score will be used to calculate the global qualification along with the other evaluation activities.

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
All forms of plagiarism in any evaluation activity and/or copying in an exam are reasons for being awarded an immediate suspend the subject.

Note: Student's assessment may experiencesome modifications depending on the restrictions to face-to-face activities enforced by health authorities.

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

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