Universitat Autònoma de Barcelona

2023/2024

Molecular Genetic Diagnostic

Code: 101973
ECTS Credits: 6

Degree	Type	Year	Semester
2500890 Genetics	OB	3	2

Contact

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Teaching groups languages

You can check it through this link. To consult the language you will need to enter the CODE of the subject. Please note that this information is provisional until 30 November 2023.

Teachers

Jordi Surralles Calonge

Massimo Bogliolo

Prerequisites

It would be appropriate to have passed and achieved the competences corresponding to the subjects: Molecular biology of eukaryotes, Cytogenetics and Human Genetics. A part of the seminars will be conducted in English and therefore it would be better for students to have a minimum level of knowledge of English in order to continue the course.

Objectives and Contextualisation

The main objectives of this subject are:

- Understand and be able to describe the main molecular analysis techniques usually used in the genetic diagnosis.
- Select with critical sense the most appropriate diagnostic techniques in each case.
- Be able to propose a project for molecular genetic diagnosis in the field of health.

Competences

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.

- Apply an entrepreneurial spirit in the area of genetics and genomics from an integrated vision of R+D+I processes.
- 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.
- 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 creativity.
- 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.
- Perform genetic diagnoses and assessments and consider the ethical and legal dilemmas.
- Produce, direct, execute and assess projects where knowledge of genetics or genomics is necessary.
- 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.
- Take the initiative and demonstrate an entrepreneurial spirit.

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. Choose the most suitable techniques to estimate epigenetic changes in each clinical case.
8. Critically select suitable techniques for genetic diagnosis
9. Describe the clinical consequences derived from epigenetic control mechanisms.
10. Describe the techniques to evaluate epigenetic changes to hereditary material.
11. Develop creativity.
12. Enumerate and describe the different techniques for analysing DNA polymorphisms that can be applied to studies of genetic variation associated to human pathologies.
13. Explain how knowledge of human genetic variation is applied to personalised medicine, pharmacogenomics and nutrigenomics.
14. Expose the ethical implications of genetic diagnosis.
15. Integrate underlying theoretical knowledge in tests used in genetic diagnosis to resolve potential sources of error in results.
16. Interpret the results obtained using techniques for the analysis of DNA polymorphisms to identify and evaluate factors of susceptibility and propensity to suffer illnesses.
17. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
18. Propose entrepreneurial genetics and genomics projects in the field of health.
19. Propose genetics or genomics projects that are applicable to the field of human health.
20. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
21. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
22. Take the initiative and demonstrate an entrepreneurial spirit.

Content

Theory

Causes of phenotypic changes
Hybridization of nucleic acids
Blotting techniques
Molecular cytogenetic techniques
Amplifiers for PCR
Analysis of amplification and PCR products
Quantitative PCR
General techniques of SNP detection
Use of microarrays
New sequencing techniques

External seminars

It will consist of seminars given by specialists on the molecular genetic diagnosis of various genetic specific diseases or groups of diseases, which includes specific clinical cases.

Seminars given by the students

The content will be agreed with the responsible professor.

Methodology

Theory classes: The student acquires their own scientific knowledge of the subject attending lectures: master classes with ICT support, which will complement with the personal study of the exposed topics. The material used in class can be found by the student in the "teaching material" tool of the Virtual Campus. These classes are conceived as a fundamentally unidirectional method of transmitting knowledge from the teacher to the student that forces them to develop autonomous learning strategies outside class.

Classes of external seminars: The knowledge acquired in the theory classes and the personal study is complemented in the seminars where specialists in the field of Clinical Genetics present the cases in which they work in which the techniques studied in the theory classes. Some of thesewill include the analysis ofspecific clinical cases. A part of the external seminars will be given in English.

Seminars given by students: Students will have to make a seminar in an oral presentation format with ICT support. These seminars will be held in groups of two students that will present cases of applications of molecular techniques in the diagnosis of genetic diseases following the model of external seminars and applying the knowledge acquired to the theory classes. The subject of the seminar will be agreed with the teacher in sufficient time. It will be valued positively that the seminars are in English.

Activities in the Virtual Campus: The student will be able to continue working with the contents of the subject through different proposals of activities that will find in the educational space of the subject in the Virtual Campus.

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.

Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
External Seminars	5	0.2	4, 6, 11, 22, 3
Seminars by the students	8	0.32	2, 11, 22, 5, 3
Theory lessons	30	1.2	2, 5, 3
Type: Supervised			
Individual tutorials	4	0.16	4, 6, 11, 22, 3
Type: Autonomous			
Bibliografic Research	16	0.64	5
Study	72	2.88	2, 5
Work at Campus Virtual	10	0.4	4, 6

Assessment

The content of the theoretical part will be evaluated by means of an objective examination. The corresponding note in this part corresponds to 50% of the final mark.

Students must do a seminar in a compulsory way and in groups of two. The evaluation of the seminars will be done through an exam that will include both the external seminars and the seminars given by the students. In addition, the teacher will also evaluate the exposure made by each student in the seminars and will positively value the student's exposure in English. The mark of this evaluation will correspond to 45% of the final mark, of which 50% corresponds to the examination of seminars and 50% to the evaluation made of the seminars given by the students.

Finally, theactive participation of the students in the various activities proposed in the teaching area of the subject on the Virtual Campus will also be evaluated (5% of the final grade).

The exhibition of the internal seminar in English will raise 0.5 on 10 the final global mark.

The student interested in improving the final grade may present a final examination of recovery of either part (theory and seminars). Introducing the recovery implies the resignation of the previously obtained note.

In order to pass it, we must draw a weighted final grade $\geq$ 4.9. The student who has not obtained this note is suspended or will be considered "non-evaluable" if it has not been submitted to any exam.

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

Single assessment

The single assessment consists of a single exam in which the contents of the entire theory and seminars program of the course will be evaluated in the form of a theoretical-practical test. The test will consist of multiple-choice questions and written answers. The grade obtained in this synthesis test will account for 72.5% of the final grade for the subject (50% for the theory part and 22.5% for the seminars part). To pass this test it is necessary to achieve a minimum score of 4,9.

Students who take advantage of the single assessment must compulsorily present the seminars in face-to-face sessions and under the same conditions as in the conventional assessment (22.5% of the final grade).

The single evaluation test will be carried out coinciding with the last partial evaluation test and the same recuperation system will be applied as for the conventional evaluation.

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Exam of Theory	50%	1.75	0.07	1, 21, 20, 2, 4, 6, 9, 10, 11, 12, 7, 13, 14, 15, 16, 17, 22, 18, 19, 8, 5, 3
Exam of seminars	22,5%	1.75	0.07	1, 21, 20, 2, 4, 6, 9, 10, 11, 12, 7, 13, 14, 15, 16, 17, 22, 18, 19, 8, 5, 3
Participation in Campus Virtual	5%	0.7	0.03	2, 4, 6, 11, 14, 22, 18, 19, 5
Seminar byt the student	22,5%	0.8	0.03	2, 4, 6, 11, 22

Bibliography

Ginsburg, G.S. and H.F. Willard (2010) Essentials of Genomic and Personalized Medicine. Elsevier (http://www.sciencedirect.com/science/book/9780123749345)
Mülhardt, C. and E.W. Beese (2007) Molecular Biology and Genomics. Elsevier (http://www.sciencedirect.com/science/book/9780120885466)
Patrinos, G.P., P.B. Danielson and W.J. Ansorg (2017) Molecular Diagnostics. Third Edition. Elsevier (http://www.sciencedirect.com/science/book/9780128029718)
Tollefsbol, T. (2011) Handbook of Epigenetics. The New Molecular and Medical Genetics. Elsevier (http://www.sciencedirect.com/science/book/9780123757098)

Software

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