

Degree programme	Type	Course
Genetics	OB	3

Contact lecturer

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Teaching staff

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Group languages

You can consult this information at the [end](#) of the document.

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

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.

Learning outcomes

- CM21 (Integrate genomic knowledge in the resolution of complex problems of human health, diagnosis and treatment.) Integrate genomic knowledge in the resolution of complex problems of human health, diagnosis and treatment.
- CM22 (Evaluate the scientific validity and ethical-legal implications of genetic testing and advanced therapies.) Evaluate the scientific validity and ethical-legal implications of genetic testing and advanced therapies.
- CM23 (Adapt communication on diagnosis, prevention and genetic counselling strategies to the needs of different audiences and social contexts.) Adapt communication on diagnosis, prevention and genetic

counselling strategies to the needs of different audiences and social contexts.

- KM16 (Describe genetic diagnosis and counseling strategies, as well as the basis of chromosomal and molecular alterations relevant to human health.) Describe genetic diagnosis and counseling strategies, as well as the basis of chromosomal and molecular alterations relevant to human health.
- SM19 (Apply molecular, cytogenetic, and computational techniques in genetic diagnosis and human forensic identification.) Apply molecular, cytogenetic, and computational techniques in genetic diagnosis and human forensic identification.
- SM20 (Interpret the genetic basis of human pathologies and the therapeutic response, integrating the gender perspective and bioethical implications. .) Interpret the genetic basis of human pathologies and the therapeutic response, integrating the gender perspective and bioethical implications. .
- SM21 (Prepare reports on scientific advances in diagnosis, genetic counselling and biotechnological therapies based on the literature review.) Prepare reports on scientific advances in diagnosis, genetic counselling and biotechnological therapies based on the literature review.

Contents

Theory

2. Causes of phenotypic changes
4. Genetic variants interpretation
6. Hybridizations of nucleic acids
8. Blotting techniques
10. Molecular cytogenetic techniques
12. Analysis of PCR amplification products
14. Quantitative PCR
16. Liquid biopsy
18. General techniques for SNP detection
20. Use of microarrays
22. New sequencing techniques applied to diagnostic

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.

Learning activities and methodology

Title	Hours	ECTS	Learning outcomes
External Seminars	5	0.2	KM16, SM19, SM21
Study	72	2.88	CM21, CM22, CM23, KM16, SM19, SM20, SM21
Work at Campus Virtual	10	0.4	SM19, SM20
Seminars by the students	8	0.32	CM21, CM22, CM23, SM20, SM21
Individual tutorials	4	0.16	CM21, CM22, CM23, KM16, SM19, SM20, SM21
Theory lessons	30	1.2	CM21, CM22, KM16, SM19, SM20
Bibliografic Research	16	0.64	CM21, CM22, CM23, KM16, SM19, SM20, SM21

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 these will include the analysis of specific 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.

Assessment

Continuous assessment activities

Title	Weight	Hours	ECTS	Learning outcomes
Exam of Theory	50%	1.75	0.07	KM16, SM19, SM20

Exam of seminars	22,5%	1.75	0.07	CM21, CM22, CM23
Seminar byt the student	22,5%	0.8	0.032	CM22, CM23, SM21
Participation in Campus Virtual	5%	0.7	0.028	CM21, SM19

1. The content of the theoretical part will be evaluated through an objective exam. The grade for this part corresponds to 50% of the final grade.
2. Students must complete a mandatory seminar in groups of two. The evaluation of the seminars will be conducted through an exam that will include both external seminars and those given by the students. Additionally, the professor will also evaluate the presentation made by each student in the seminars and will positively value students who give their presentation in English. The grade for this evaluation will correspond to 45% of the final grade, of which 50% corresponds to the seminar exam and 50% to the evaluation of the seminars given by the students.
3. Finally, students' active participation in the various activities proposed in the course's space on the Virtual Campus will also be evaluated (5% of the final grade).
4. Presenting the internal seminar in English will increase the final overall grade by 0.5 out of 10.
5. At the end of the presentations, students will vote for the best one. The winning group will be awarded an additional 0.5 points on their seminar evaluation grade. Only students who have attended 100% of the seminars in person will be able to vote and be eligible for the award.

Students interested in improving their final grade have the option to take a re-examination for both the theory and seminar components. Opting for the re-examination waives any previously obtained grade.

To pass, a weighted final grade of at least 4.9 must be achieved. A student who fails to obtain this grade will be marked as \"failed\" or \"non-evaluable\" if no exams were taken.

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 weighting 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.

Use of AI

For this course, the use of artificial intelligence (AI) technologies is permitted exclusively for support tasks, such as bibliographic or information searches, text correction, or translations.

For submissions on the Virtual Campus (CV) and in seminars, the student must clearly identify which parts have been generated with this technology, specify the tools used, and include a critical reflection on how they have influenced the process and the final outcome of the activity.

The non-transparent use of AI in this assessable activity will be considered a lack of academic honesty and may lead to a partial or total penalty in the activity's grade, or more significant sanctions in serious cases.

Irregularities during assessment activities

Any irregularity committed during an assessment activity (academic fraud, plagiarism, or the inappropriate use of AI, unless such use is expressly authorized in the course syllabus) that could lead to a significant variation in the grade will result in a grade of 0 for that activity. If the course syllabus stipulates that obtaining a minimum grade in this assessment activity is a mandatory requirement to pass the course, or if multiple irregularities occur in the assessment activities of the same course, the final grade for the course will be 0. In addition to this, disciplinary proceedings may be initiated against any student who commits such irregularities.

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

No aplica

Course groups and languages

The information provided is provisional until November 30. After this date, you will be able to consult the language of each group through this [link](#). To access the information, you will need to enter the course CODE

Type of teaching	Group	Language	Semester	Shift
(TE) Theory	63	Spanish	second semester	morning-mixed
(SEM) Seminars	631	English	second semester	morning-mixed
(SEM) Seminars	632	English	second semester	morning-mixed