

Degree	Type	Year
2500890 Genetics	OB	3

Contact

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Teaching groups languages

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Prerequisites

To ensure proper monitoring of the subject and the achievement of the learning outcomes, it is recommended:

1. Familiarize and understand the basic foundations of the first-year subjects: "Cellular Biology and Histology" and "Genetics."
2. Familiarize and understand the basic foundations of the second-year subjects: "Cytogenetics."
3. Acquire knowledge of the techniques used in these disciplines, as many of them will be introduced throughout the course and assumed to be known.

Objectives and Contextualisation

Sexual reproduction in most species is associated with sexual dimorphism and the presence of chromosomes that determine sex. Sexual dimorphism is achieved through the involvement of specific genes involved in differential sexual development. Mutations in these genes condition sexual differentiation and, therefore, the fertility of affected individuals. On the other hand, gametogenesis is a complex and highly regulated process. Dysfunctions or abnormalities affecting one or more stages involved in the formation of sperm and oocytes can impair the reproductive capacity of affected individuals. To date, the relationship between various genotypic alterations and their effect on the reproductive capacity of individuals has been established. Generally, the manifestation of genetically originated infertility is associated with a significant reduction in the number of produced gametes, abnormalities in embryonic development, or spontaneous abortions.

In this context, the objectives of the subject will focus on:

1. Establishing the genetic causes that condition reproduction in humans.
2. Reviewing genetic analysis techniques aimed at the characterization of gametes and preimplantation embryos.
3. Determining the risk of transmission of genetically caused infertility.
4. Establishing the foundations for reproductive genetic counseling.

Competences

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
- Apply knowledge of theory to practice.
- Assume ethical commitment
- Define mutation and its types, and determine the levels of genic, chromosomal and genomic damage in the hereditary material of any species, both spontaneous and induced, and evaluate the consequences.
- 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 experiments and interpret the results.
- Develop self-directed learning.
- 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.
- 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. Apply the basic principles of bioethics.
4. Assess the implication of genetic anomalies as a cause of infertility.
5. Assume ethical commitment
6. Describe the basic genetic techniques for the study and prevention of sterility and infertility.
7. Describe the genetic bases of the determination and differentiation of human gender.
8. Describe the genetic basis and control of human gametogenesis.
9. Describe the structure and variation of the human genome from a functional and evolutionary perspective.
10. Design experiments and interpret the results.
11. Develop self-directed learning.
12. Explain cancer as an error in the control mechanisms of genic expression.
13. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
14. Perform pre-conceptual genetic assessment taking into account its ethical and legal implications.
15. Recognise the genetic anomalies of spermatogenesis and oogenesis related with a sterility phenotype.
16. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
17. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.

Content

SECTION I: GENETIC BASIS OF REPRODUCTION

Topic 1. Sex determination and differentiation in humans

Topic 2. Genetic control of human gametogenesis

SECTION II: GENETIC BASIS OF HUMAN INFERTILITY

Topic 3. Genetic basis of male infertility

Topic 4. Genetic basis of female infertility

SECTION III: GENETIC DIAGNOSIS AND ASSISTED HUMAN REPRODUCTION

Topic 5. Introduction to human-assisted reproduction techniques

Topic 6. Genetic studies in infertile couples

Topic 7. Preimplantational genetic diagnosis

Topic 8. Preconceptional genetic studies in gamete donors and couples with a reproductive desire

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Problem solving	4	0.16	2, 5, 10
Theory classes	22	0.88	4, 6, 7, 8, 14, 15
Type: Autonomous			
Individual study	27	1.08	11
Problem solving	16	0.64	2, 5, 10, 11

Theory Classes

The content of the theory program will be explained by the teacher in lessons of 50 minutes with audiovisual support, encouraging active student participation. The tables, figures, and graphs used in the sessions will be available in PDF format in the Moodle classroom of the subject. Students will also have access through this platform to recommended videos, animations, and websites, and will receive detailed bibliography for each topic, which they should consult to consolidate the theoretical classes and for personal study of the explained topics.

Problem Classes

Students will be organized into groups of four and must attend the corresponding sessions in the group assigned by the degree coordination. Each student will complete 4 sessions of 50 minutes during the course. The organization in the classroom will be as follows:

1. Students will have a list of problems to solve in advance.
2. For each scheduled session, students will work on 3 to 4 problems and prepare a response dossier.
3. Before each session, each team will submit the response dossier (a group submission) in the designated space in the Moodle classroom of the subject.
4. The problems will be solved in the classroom on the assigned day according to the subject's calendar. The teacher will choose a student who will perform an oral presentation to the rest of the students.

5. The problem-solving and presentation will be evaluated by the teacher, and the grade obtained will be applied to all members of the team to which the student belongs.
6. At the end of each session, the teacher will select and grade a problem from the dossier. The grade obtained will be applied to all members of the work team and will contribute to the final grade of this activity.

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
Problem-solving	20	2	0.08	1, 2, 11, 13, 16, 17
Written examination I	40	2	0.08	1, 3, 4, 6, 7, 8, 9, 11, 12, 13, 15, 16, 17
Written examination II	40	2	0.08	1, 2, 4, 5, 6, 10, 11, 13, 14, 15, 16, 17

To pass the subject, it will be essential to obtain a final grade equal to or higher than 5 points out of 10 based on the contributions from different evaluation activities. Additionally, students must achieve a minimum grade in the average of the two written exams equal to or higher than 4 points out of 10.

Written Exam I and Written Exam II (individual evaluation)

Throughout the semester, two written tests (consult the subject's schedule) on the theoretical contents of the subject will be conducted, which students must individually respond to. The tests will consist of multiple-choice questions aimed at assessing the mastery of concepts and knowledge covered in class and verifying their correct application and interrelation. Each of the tests will have a weight of 40% on the final grade of the subject. Students must obtain a minimum grade in the average of both tests of 4 points (out of 10) in order to pass the subject.

Problem Solving (group evaluation)

The grade for this part will be obtained through the average of the scores obtained by each group of students throughout the course (one problem per dossier) and through the oral resolution in class. The teacher will ensure that each group presents at least one exposition during the course. A problem not submitted or not solved in class will be scored as zero in the group's average grade calculation. The evaluation of the problems will take into account the correctness of the answer, the formulation, and the interpretation of the results. The final grade will be shared by all members of each group and will account for 20% of the final grade.

Recovery Exam

There will be a recovery exam for those students who have not achieved the required grade in the partial exams evaluating the theoretical contents (average of 4 out of 10) or who have not reached the minimum grade to pass the subject (5 points out of 10). To participate in the recovery exam, students must have been previously evaluated in a set of activities that account for at least two-thirds of the total grade of the subject. Therefore, students will receive a "Not evaluable" grade when the weight of the evaluation activities is less than 67% of the final grade. To include the grade obtained in the recovery exam in the final grade of the subject, it will be necessary to achieve a grade of 4 out of 10 in that exam.

Single Evaluation

Students will have the option to be evaluated on the theoretical contents of the subject through a single written exam (individual evaluation), which will account for 80% of the final grade. The remaining 20% will be based on problem-solving, which will be continuously evaluated throughout the course (see previous section). This exam will be of the same type as the two exams scheduled for the continuous evaluation of the theoretical contents. It will take place on the same date specified in the calendar for Written Exam II, and the same recovery system as the continuous evaluation will be applied.

Students who choose this modality must inform the responsible professor of the subject before the first working day of October.

Bibliography

Bajo JM, B. Coroleu B. (Eds.) Fundamentos de Reproducción. Editorial Panamericana. Madrid. 2009.

Elder K., Dale B. In vitro fertilization. (3rd edition). Cambridge University Press. New York. 2011.

Fauser B.C.J.M. (Ed.). Molecular Biology in Reproductive Medicine. The Parthenon Publishing Group. New York. 1999

Gardner D.K. et al. (Eds.). Textbook of assisted Reproductive Techniques. Martin Dunitz Pub. Hampshire. 2001.

Harper J. (Ed.) Preimplantation Genetic Diagnosis. (2nd Edition). Cambridge University Press. New York (USA).2009.

Johnson M.H. and Everitt B.J. (Eds.) Essential Reproduction. 5th Edition. Blackwell Science. Oxford. 2005.

Matorras R, Hernández J. (Eds.). Estudio y tratamiento de la pareja estéril. Adalia. Madrid. 2007.

Specific review articles will be recommended during the course.

Software

Not applicable

Language list

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