

Genetics and Reproduction

Code: 101891
ECTS Credits: 3

Degree	Type	Year	Semester
2501230 Biomedical Sciences	OT	4	1

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

Prerequisites

To ensure the achievement of the learning aims, it is highly recommended:

1. An appropriate knowledge of the subjects "Cell Biology and Histology", "Genetics" and "Cytogenetics", completed in previous courses of the degree
2. An appropriate knowledge of the techniques used in these disciplines
3. Basic knowledge of basic computer tools
4. Good command in English reading

Objectives and Contextualisation

Sexual reproduction in most species is associated with sex dimorphism and the presence of chromosomes that determine sex. Sex dimorphism is achieved through the participation of specific genes involved in a differential sex development. Mutations in these genes influence normal sex differentiation and therefore the fertility of affected individuals. On the other hand, gametogenesis is a complex and highly regulated process whose alterations can lead to infertility.

The genetic contribution to infertility is difficult to assess. Up to date, it has been established the relationship between several genotype alterations and their effect on fertility. However, except for few diseases (for example cystic fibrosis), these patients do not exhibit any relevant phenotypic traits. In general, the manifestation of infertility of genetic origin is related to a significant reduction in the number of gametes produced, anomalies in embryo development and spontaneous abortions.

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

1. To establish the genetic causes of human infertility.
2. To provide updated knowledge about the methodologies for the genetic analysis of gametes and embryos.
3. To determine the risk of transmission of those cases of infertility caused by genetic alterations.
4. To establish the basis for reproductive genetic counseling.

Competences

- Display knowledge of the bases and elements applicable to the development and validation of diagnostic and therapeutic techniques.
- Display knowledge of the basic life processes on several levels of organisation: molecular, cellular, tissues, organs, individual and populations.
- 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.

Learning Outcomes

1. Correctly use the terminology of genetics and its text and reference books
2. Describe and understand the genetic bases and control of human gametogenesis.
3. Describe and understand the genetic bases of sex determination and differentiation in humans.
4. Describe the genetic techniques for the study and prevention of sterility and infertility.
5. Evaluate the involvement of genetic anomalies as a cause of infertility.
6. Interpret scientific publications and solve problems and typical cases in the area of cytogenetics.
7. Provide pre-conceptional genetic counselling, taking ethical and legal factors into account.
8. Recognise genetic anomalies in spermatogenesis and oogenesis related to a phenotype of sterility.
9. Understand and describe the structure, morphology and dynamics of eukaryote chromosomes in the different stages of the cell cycle.

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

Methodology

Theory classes

The content of the theory program will be delivered by the teacher as lectures, with audio-visual support and encouraging the active participation of students, in 22 sessions of 50 minutes.

Tables, figures, and graphs used in the sessions will be available in pdf format on the corresponding Moodle class. Students will also have access through this platform to videos, animations, and recommended websites.

Students will receive detailed bibliography to consolidate the topics explained in theory classes.

Problem-solving classes

The aims of the sessions are:

1. To initiate the students in the resolution of representative experiments that clearly illustrate new advances in the field.
2. To consolidate the concepts developed in theory classes, as well as evaluate the implications that arise from them.
3. To initiate the students in the scientific method, working with the learning objectives especially related to reasoning, critical judgment, and communicative skills.

In these sessions, the students will be divided into two groups and will work in small groups (four to six people) to resolve problems and cases proposed. Students must attend the sessions corresponding to the assigned group. Along the course, each student will complete four sessions of 50 minutes. The organization of the task in the classroom will be done as follows:

1. Students will have a list of problems to be solved in advance. For each of the scheduled sessions, students must solve 3-4 problems and prepare a dossier response according to the instructions given by the teacher.
2. Before each discussion session, each team will deliver a response dossier (one team delivery). Problems will be further discussed and corrected in the classroom, with the active participation of the students. A member of the team will be randomly chosen by the teacher for an oral presentation to the rest of the students. The resolution of the problem and the presentation will be evaluated by the teacher and the qualification obtained will be applicable to all the members of the team to which the student belongs.
3. At the end of each session, the teacher will choose a problem to be evaluated and the qualification obtained will be applicable to all members of the work team.

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			
Lectures	22	0.88	9, 3, 2, 4, 7, 8, 1, 5
Problem sessions	4	0.16	3, 2, 4, 6, 7, 8, 1, 5
Type: Autonomous			
Problem resolution	16	0.64	6, 7, 8, 1, 5
Study and self learning concepts	27	1.08	9, 3, 2, 4, 6, 7, 8, 1, 5

Assessment

Students must get a final mark equal to or greater than 5 points (out of 10) to pass the subject

Written exams I and II (individual assessment)

Two written multiple-choice tests will be scheduled to evaluate the degree of achievement of the concepts related to the subject. Students will have to answer it individually.

Each of the tests will have a weight of 40% on the final qualification of the subject. To pass the subject, students must obtain a minimum mark of 4 points (out of 10) of the mean of the two test exams.

Problem-solving (group assessment)

The qualification will be obtained by the mean of the marks obtained in the reports delivered by each team along the course (one problem per report) and in the classroom oral presentations.

The teacher will supervise that at least one presentation was done by each team. A problem not delivered or not resolved in the class will receive a mark of zero. Qualifications will be assigned according to the answers given in the reports, the approach of the solutions, and the interpretation of the results.

The final qualification obtained will be the same for all the members of the team and will weigh 20% of the final qualification of the subject.

Retake exam

There will be a retake exam for those students who have not passed the partial exams (mean of 4 points out of 10) or who have not achieved the minimum mark required to pass the subject (5 points out of 10).

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 "Not Evaluable" if the weighting of all conducted evaluation activities is less than 67% of the final score.

Students must get at least 4 points (out of 10) in the qualification obtained in the retake exam to make an average with the solving problems marks. The methodology of the retake exam may be different from that used in previous evaluations.

Exam review (contesting of the grade)

The review of the exams will be done on the dates scheduled by the teachers.

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Problem resolution	20	2	0.08	3, 2, 4, 6, 7, 8, 1, 5
Written examination I	40	2	0.08	9, 3, 2, 6, 8, 1
Written examination II	40	2	0.08	4, 7, 8, 1, 5

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

To follow the course, students must regularly consult documents in pdf format.