

**Genetics and Reproduction**

Code: 104120  
ECTS Credits: 3

Degree	Type	Year	Semester
2500890 Genetics	OB	3	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

**Teachers**

Joan Blanco Rodríguez

**Prerequisites**

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

1. An appropriate knowledge of subjects completed in previous courses: "Cell Biology and Histology", "Genetics" and "Cytogenetics".
3. An appropriate knowledge of the techniques used in these disciplines.
4. Basic knowledge of the use of computer tools (Internet, Powerpoint, and word processors)
5. A good English reading comprehension

**Objectives and Contextualisation**

Sexual reproduction in most species is associated to 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 influences normal sex differentiation and therefore reproduction. On the other hand, gametogenesis are complex and highly regulated processes. Dysfunctions or anomalies that affect one or more stages involved in the formation of sperm or oocytes can impair the reproductive capacity of the individuals affected.

The genetic contribution to fertility problems is difficult to assess. Up to date, it has been established the relationship between several genotype alterations and their effect on the reproductive capacity of the affected individuals. 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 or spontaneous abortions.

In this context, the objectives will focus on:

1. To establish genetic causes conditioning reproduction in humans.

2. To provide updated knowledge about assisted reproductive techniques and the applications of in vitro handling of gametes and embryos.
3. To determine the risk of transmission to the offspring.
4. To establish the basis for reproductive genetic counselling

## **Skills**

- 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.
- 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.

## **Learning outcomes**

1. Apply knowledge of theory to practice.
2. Apply the basic principles of bioethics.
3. Assess the implication of genetic anomalies as a cause of infertility.
4. Assume ethical commitment
5. Describe the basic genetic techniques for the study and prevention of sterility and infertility.
6. Describe the genetic bases of the determination and differentiation of human gender.
7. Describe the genetic basis and control of human gametogenesis.
8. Describe the structure and variation of the human genome from a functional and evolutionary perspective.
9. Design experiments and interpret the results.
10. Develop self-directed learning.
11. Explain cancer as an error in the control mechanisms of genic expression.
12. Perform pre-conceptual genetic assessment taking into account its ethical and legal implications.
13. Recognise the genetic anomalies of spermatogenesis and oogenesis related with a sterility phenotype.

## **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 BASES OF 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. Assisted Reproduction Techniques (TRAs)

Topic 6. Risks of Assisted Reproduction Techniques

Topic 7. Genetic analysis of gametes

Topic 8. Preimplantation genetic diagnosis

Topic 9. Prenatal genetic diagnosis

Topic 10. Reproductive genetic counseling

## Methodology

### Theory classes

The content of the theory program will be presented by the teacher as lectures, with audio-visual support and encourage 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 UAB-Virtual Campus. Students 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 aim of the sessions is

- To initiate the student in the resolution of representative experiments that clearly illustrate new advances in the field
- To consolidate the concepts developed in theory classes, as well as evaluate the implications that arise from them.
- 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. Student must attend the sessions corresponding to the assigned group. Each student will complete during the course 4 sessions of 50 minutes.

1. Students will have the 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, following the instructions of the teacher, each team will deliver a response dossier (one team delivery). Problems will be further discussed and corrected in the classroom, with active participation of the students. A member of the team will be chosen randomly by the teacher for an oral presentation to the rest of 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 randomly a problem to be evaluated based on the response dossier. The qualification obtained will be applicable to all members of the work team.

## Activities

Title	Hours	ECTS	Learning outcomes
Type: Directed			

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Problem solving	4	0.16	1, 4, 9, 12, 13, 3
Theory classes	22	0.88	2, 5, 6, 7, 12, 13, 3
<b>Type: Autonomous</b>			
Individual study	27	1.08	2, 5, 6, 7, 10, 12, 13, 3
Problem solving	16	0.64	1, 2, 4, 10, 9, 12, 13, 3

## Evaluation

To pass the subject, the student must get at least 5 points (out of 10).

### Evaluation activities

#### **Written examination I and II (individual assessment)**

Two written multiple-choice test will be scheduled to evaluate the degree of achievement of theory concepts of the subject. Students will have to answer 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 arithmetic mean of the two test exams.

#### **Problem solving (group assessment)**

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

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 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 weight to 20% of the final qualification of the subject.

#### **Retake exam**

There will be a retake exam of the subject for those students who have not passed the partial theory examinations (mean of 4 points out of 10) or who have not achieved the minimum marks 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 "No Avaluable" if the weighting of all conducted evaluation activities is less than 67% of the final score.

The student must get at least 4 points (out of 10) in the qualification obtained in the retake exam to make average

the solving problems marks.

The methodology of the retake examination may be different from that used in previous evaluations.

#### **Examination revision**

The revision of exams will be by appointment according to the dates scheduled by the teachers

## Evaluation activities

Title	Weighting	Hours	ECTS	Learning outcomes
Problem-solving	20	2	0.08	2, 5, 10, 12, 13, 3
Written examination I	40	2	0.08	5, 8, 6, 7, 10, 11, 13, 3
Written examination II	40	2	0.08	1, 2, 4, 5, 10, 9, 12, 13, 3

## 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 bibliography will be indicated during the course**