Universitat Autònoma de Barcelona

2020/2021

Genetics and Reproduction

Code: 101891
ECTS Credits: 3

Degree	Type	Year	Semester
2501230 Biomedical Sciences	OT	4	0

The proposed teaching and assessment methodology that appear in the guide may be subject to changes as a result of the restrictions to face-to-face class attendance imposed by the health authorities.

Contact

Name: Francesca Vidal Domínguez
Email: Francesca.Vidal@uab.cat

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

There are no prerequisites for taking this course, however to ensure the achievement of the learning aims, it is recommended to:

1. Have an appropriate knowledge of subjects completed in previous courses: "Cell Biology and Histology", "Genetics" and "Cytogenetics".
3. Have an appropriate knowledge of the techniques used in these disciplines.
4. Have a basic knowledge of the use of computer tools (Internet, Powerpoint, and word processors)

5. Have 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 stablished 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 stablish 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

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 ovogenesis 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 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

Unless the requirements enforced by the health authorities demand a prioritization or reduction of these contents.

Methodology

Theory classes

The content of the theory program will be presented 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 UAB-Virtual Campus. 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 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 studets 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 correctedin the classroom, with activeparticipation of the students. A member of theteam will bechosen randomly by the teacher for an oral presentation to the rest of students. The resolution of the problem and the presentationwill 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.

The proposed teaching methodology may experience some modifications depending on the restrictions to face-to-face activities enforced by health authorities

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 ressolution	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

To pass the subject, the student must get a final grade equal or greater than 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.

A minimum score of 4 out of 10 in this examination is required to pass the course.

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

Exam revision

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

Student's assessment may experience some modifications depending on the restrictions to face-to-face activities enforced by health authorities

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Problem ressolution	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.

Reviews and updated papers will be recommended during the course