

2021/2022

# Case Studies in Cytogenerics and Reproduction Biology

Code: 42947 ECTS Credits: 6

Degree	Туре	Year	Semester
4313782 Cytogenetics and Reproductive Biology	ОВ	0	2

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: Vicenç Català Cahís

Email: Vicenc.Catala@uab.cat

# **Teachers**

Joan Francesc Barquinero Estruch

Vicenç Català Cahís

Zaida Sarrate Navas

Alberto Plaja Rustein

Keyvan Torabi Asensio

## **External teachers**

Anna Rabanal

Carles Garrido

Carmen Marquez

Elisabeth Clua

Emma Triviño

Esther Gean

Francesc Solé

Lluch Coll

Maria Carme Pons

Mark Grossmann

Marta Moragas

Olga Martinez Pasarell

Rafel Buscà

Silvia Mateo

Susana Egozcue

Vincenzo Cirigliano

# **Use of Languages**

Principal working language: catalan (cat)

# **Prerequisites**

There are no specific prerequisites.

However, in order to ensure the correct follow-up of the sessions, it is recommended to have a good knowledge

It is recommended to have a good knowledge of English.

# **Objectives and Contextualisation**

To provide to the students a close interaction with experts of the field and insights for their professional carrer in t to the students.

# **Competences**

- Apply knowledge of theory in both research and clinical care contexts.
- Apply the scientific method and critical reasoning to problem solving.
- Communicate and justify conclusions clearly and unambiguously to both specialist and non-specialist audiences.
- Continue the learning process, to a large extent autonomously.
- Design experiments, analyse data and interpret findings.
- Identify the ethical dilemmas and apply current laws governing the area of knowledge of the master's degree.
- Integrate knowledge and use it to make judgements in complex situations, with incomplete information, while keeping in mind social and ethical responsibilities.
- Interpret, resolve and report on clinical cases or scientific findings in the area of the master's degree.
- Organise and manage research laboratories and clinical laboratories in the area of knowledge of the master's degree.
- Show an ability to work in teams and interact with professionals from other specialist areas.
- Solve problems in new or little-known situations within broader (or multidisciplinary) contexts related to the field of study.
- Use acquired knowledge as a basis for originality in the application of ideas, often in a research context.
- Use and manage bibliography or ICT resources in the master's programme, in one's first language and in English.

• Use creative, organisational and analytic skills when taking decisions.

# **Learning Outcomes**

- 1. Apply current laws on clinical practice in cytogenetics and reproductive biology.
- 2. Apply current laws on genetic counselling.
- 3. Apply current laws on laboratory management.
- 4. Apply knowledge of theory in both research and clinical care contexts.
- 5. Apply the scientific method and critical reasoning to problem solving.
- 6. Assess the prognosis factor for certain cancers, based on the genetic alterations diagnosed, and contribute to genetic counselling.
- Communicate and justify conclusions clearly and unambiguously to both specialist and non-specialist audiences.
- 8. Continue the learning process, to a large extent autonomously.
- 9. Design experiments, analyse data and interpret findings.
- Evaluate the risk of affected offspring in carrier individuals and contribute to reproductive genetic counselling.
- 11. Evaluate the risk of chromosomal or molecular alterations and contribute to genetic counselling.
- 12. Identify and diagnose chromosomal and molecular alterations involved in human pathologies.
- 13. Identify and diagnose genetic anomalies involved in human sterility/infertility.
- 14. Identify and solve safety problems and specific laboratory infrastructures.
- 15. Identify the ethical dilemmas associated with clinical practice in cytogenetics and reproductive biology.
- 16. Integrate knowledge and use it to make judgements in complex situations, with incomplete information, while keeping in mind social and ethical responsibilities.
- 17. Show an ability to work in teams and interact with professionals from other specialist areas.
- 18. Solve problems in new or little-known situations within broader (or multidisciplinary) contexts related to the field of study.
- 19. Use acquired knowledge as a basis for originality in the application of ideas, often in a research context.
- 20. Use and manage bibliography or ICT resources in the master's programme, in one's first language and in English.
- 21. Use creative, organisational and analytic skills when taking decisions.

# Content

Topics will be presented as case reports by professionals from research centers, assisted reproduction centers and genetic centers. The syllabus will be developed in the form of clinical cases selected by the lecturers. The coordinator of the subject will evaluate the appropriateness of the cases.

# Topic 1: Cytogenetic cases\*

Practical cases of prenatal, postnatal genetic diagnosis, constitutional and cancer cytogenetics will be studied. The most appropriate diagnostic methodologies for each case will be addressed together with the interpretation of the results and decisions taking.

Cases will include: prenatal diagnosis of aneuplodies, prenatal diagnosis of monogenic diseases, structural chromosomal abnormalities, chromosomal variants, disorders caused by deletions and cancer genetics and cytogenetics. Hypothesis will be established based on the results and the most appropriate interventions will be discussed for each situation.

# Topic 2: Biology of reproduction\*

Clinical of genetic diagnosis of female and male infertility, assisted reproduction treatments, donation of gametes and pre-embryos, cryopreservation of gametes and embryos and preimplantation genetic diagnosis cases will be analyzed. Hypothesis will be established based on the results and the most appropriate interventions will be discussed for each situation

Cases will include: application of assisted reproduction techniques in female factors, application of reproductive techniques assisted in male factors, ovarian hyperstimulation syndrome (OHS), recurrent pregnancy loss, in vitro embryo culture, embryo morphokinetics, preimplantation genetic diagnosis.

Note: \*

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

# Methodology

Resolution of clinical cases presented by professionals.

Preparation and presentation of cases and assumptions by the students.

Seminars.

Debates

Autonomous study

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

"Teachers should allocate approximately 15 minutes of some class to allow their students to answer the surveys of evaluation of teaching performance and evaluation of the subject or module"

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

Resolution of clinical cases in class	44	1.76	
Type: Supervised			
Presentation of cases and assumptions by the students. Seminars. Clinical Simulation	20	0.8	5, 4, 1, 2, 3, 10, 11, 17, 9, 21, 15, 12, 13, 14, 16, 18, 7, 8, 19, 20, 6
Type: Autonomous			
Preparation follow-up cases. Preparation and presentation of cases and assumptions. Preparation of seminars and debates		3.04	5, 4, 1, 2, 3, 10, 11, 17, 9, 21, 15, 12, 13, 14, 16, 18, 7, 8, 19, 20, 6

#### **Assessment**

It will be essential to pass the subject to obtain a final mark equal to or greater than 5 points (out of 10).

#### Evaluation activities:

- 1. A written exam will be carried out at the end of the programmed teaching (see the date in the course calendar) that the students will have to answer individually. This test will consist of a multiple choice test questions. The objective is to evaluate the achievement of the concepts and the knowledges treated in class and verify the correction at the time to apply and to relate them. A clinical visit with a simulated patient may be included. This test will weight a 40% on the final mark of the subject. Students must obtain a minimum score of 3.5 points (out of 10) in this exam to be able to pass the subject.
- 2. Students will be required to individually prepare a case study that will be handed in to the responsible teachers on the proposed date. Each student will have to make the presentation and oral defense of the supposed case delivered. The evaluation of this activity will be carried out taking into account the elaboration and approach of the case and the presentation / oral defense of the case. Participation in the debates This activity will have a weight of 60% on the final note of the asignatura \*.

Taking into account that the attendance to the classes is obligatory, the following correction will be applied to determine the final note of the subject:

100-80% attendance: The final grade will be 100% of the score obtained from the assessment activities  $\le$ 80 -  $\ge$ 50% attendance: The final grade will be 75% of the score obtained from the assessment activities <50% attendance: The final grade will be 50% of the score obtained from the assessment activities

#### Retake test:

There will be a review of the subject's for those students who have not passed the minimum grade required in the theoretical exam (3.5 out of 10) or who have not obtained the minimum grade to pass the subject (5 points on 10).

#### Exam reviews

The exam reviews will be with a concerted appointment on the dates proposed by the responsible professors

Note: \*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
Preparation of an assumed case	40%	7	0.28	5, 4, 1, 2, 10, 11, 17, 9, 21, 15, 12, 13, 14,

16, 18, 7, 8, 19, 20, 6

Presentation of an alleged case. Clinical case simulation.	20%	1	0.04	5, 4, 1, 2, 3, 10, 11, 17, 9, 21, 15, 12, 13, 14, 16, 18, 7, 8, 19, 20, 6
Written tests	40%	2	0.08	1, 2, 3, 10, 11, 15, 12, 13, 14, 18, 6

# Bibliography

It will be recommended during the sessions.

Each specialist will provide a list of bibliography.

# **Software**

There is no specific software for this course