

Degree programme	Type	Course
Cytogenetics and Reproductive Biology	OB	1

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Group languages

You can consult this information at the [end](#) of the document.

Prerequisites

There are no specific prerequisites for enrolling in this course.

However, to ensure adequate follow-up and understanding of the practical case studies, students are expected to have assimilated the knowledge acquired in the theoretical and practical courses previously completed within the Master's programme.

Students are also encouraged to have a working knowledge of English, as information sources and recommended bibliography are frequently provided in this language.

Objectives

To provide the students a close interaction with experts of the field and insights for their professional career in the future. To bring professional opportunities to the students.

Learning outcomes

- CA04 (Interpret clinical cases in the field of clinical cytogenetics and reproductive biology, integrating genetic, cytogenetic and clinical results to formulate a diagnosis or etiological hypothesis.) Interpret clinical cases in the field of clinical cytogenetics and reproductive biology, integrating genetic, cytogenetic and clinical results to formulate a diagnosis or etiological hypothesis.
- CA05 (Examine, using real cases from cytogenetics or reproductive biology, the procedures and criteria relating to quality, safety and traceability applied in specialist laboratories, identifying best practices and key aspects of their operation.) Examine, using real cases from cytogenetics or reproductive biology, the procedures and criteria relating to quality, safety and traceability applied in specialist laboratories, identifying best practices and key aspects of their operation.
- CA06 (Assess, with ethical and professional responsibility, the validity of a diagnosis and formulate a prognosis based on a case study involving cytogenetic abnormalities, cancer or infertility.) Assess, with ethical and professional responsibility, the validity of a diagnosis and formulate a prognosis based on a case study involving cytogenetic abnormalities, cancer or infertility.
- KA09 (Identify the origin of human pathologies associated with genetic abnormalities from real example cases.) Identify the origin of human pathologies associated with genetic abnormalities from real example cases.
- KA10 (Explain the genetic and epigenetic changes involved in the aetiology and progression of diseases and constitutional malformations.) Explain the genetic and epigenetic changes involved in the aetiology and progression of diseases and constitutional malformations.
- KA11 (Relate the origin, causes and treatments of human infertility based on clinical cases.) Relate the origin, causes and treatments of human infertility based on clinical cases.
- SA06 (Apply analysis and diagnosis protocols in patients with constitutional chromosomal abnormalities, cancer and infertility.) Apply analysis and diagnosis protocols in patients with constitutional chromosomal abnormalities, cancer and infertility.
- SA07 (Solve real clinical cases in patients with genetic diseases and/or infertility problems, integrating clinical and genetic information to formulate diagnostic hypotheses.) Solve real clinical cases in patients with genetic diseases and/or infertility problems, integrating clinical and genetic information to formulate diagnostic hypotheses.

Contents

The course will be taught by professionals from research centers, assisted reproduction centers, and genetic

diagnostic laboratories, both national and international.

The syllabus will be developed through clinical case studies selected by the participating faculty members.

The module coordination team will supervise the suitability of the proposed cases, ensuring a balanced representation of the two master's specializations (cytogenetics and reproductive biology).

Block 1: Case Studies in Cytogenetics

Practical cases in prenatal and postnatal genetic diagnosis, constitutional cytogenetics, and the cytogenetics of solid tumors and oncohematological malignancies will be presented. The most appropriate diagnostic methodologies for each case, the integration and interpretation of results, and decision-making processes will be discussed.

Cases will include: prenatal diagnosis of aneuploidies, prenatal diagnosis of monogenic diseases, structural chromosomal abnormalities, chromosomal variants, disorders caused by deletions/microduplications, and cancer-associated chromosomal abnormalities.

Block 2: Case Studies in Reproductive Biology

Practical cases involving the genetic diagnosis of female and male infertility, assisted reproductive treatments, gamete and pre-embryo donation, cryopreservation of gametes and pre-embryos, and preimplantation genetic testing will be studied. Appropriate diagnostic methodologies will be discussed, hypotheses will be formulated based on the results obtained, and the most suitable interventions for each case will be evaluated. Cases will include: application of assisted reproduction techniques in female-factor infertility, application of assisted reproduction techniques in male-factor infertility, ovarian hyperstimulation syndrome (OHSS), recurrent miscarriage, monitoring of in vitro embryo culture through morphokinetic parameters, preimplantation genetic testing for chromosomal abnormalities, and preimplantation genetic testing for monogenic disorders.

A professional career guidance session and the development of an individualized career plan will also be included.

Use of Artificial Intelligence: For this course, the use of Artificial Intelligence (AI) technologies is permitted exclusively for literature and information searches, as well as for text correction and improvement. The use of AI for the de novo generation of texts or presentations is not permitted under any circumstances. Students must clearly identify which parts of their work have been generated using AI technologies, specify the tools used, and include a critical reflection on how these tools influenced both the process and the final outcome of the activity. Lack of transparency regarding the use of AI in this assessed activity will be considered a breach of academic integrity and may result in partial or total penalties in the activity grade, or more severe disciplinary measures in cases of serious misconduct.

Irregularities in assessment activities: Any irregularity committed in an assessment activity (academic fraud, plagiarism, or improper use of AI, unless such use is explicitly authorized in the course syllabus) that may lead to a significant alteration of the grade will result in that assessment activity being graded as 0. If the course syllabus establishes that obtaining a minimum grade in that assessment activity is an essential requirement for

passing the course, or if multiple irregularities occur in the assessment activities of the same course, the final grade for the course will be 0. Furthermore, disciplinary proceedings may be initiated against any student involved in such irregularities.

Learning activities and methodology

Title	Hours	ECTS	Learning outcomes
Resolution of clinical cases in class	44	1.76	
Presentation of cases and assumptions by the students. Seminars. Clinical Simulation	20	0.8	
Preparation follow-up cases. Preparation and presentation of cases and assumptions. Preparation of seminars and debates	76	3.04	

The methodology followed in the sessions will consist of:

- Lectures and presentations delivered by professionals.
- Resolution of clinical cases proposed by the professionals.
- Preparation and presentation of cases and scenarios by the students.
- Discussion of the cases presented.
- Reflection on career development planning.
- Independent study.

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

Continuous assessment activities

Title	Weight	Hours	ECTS	Learning outcomes
Preparation and presentation of a case study	40%	6	0.24	CA05, CA06, KA10, KA11, SA06, SA07
Test exam	30%	2	0.08	CA04, CA05, CA06, KA09, KA10, KA11, SA06, SA07
Career plan	10%	1	0.04	CA06, KA11
Defense of a case study	20%	1	0.04	CA04, CA06, KA10, KA11, SA06, SA07

This module does not include a single assessment system.

To pass the course, it is mandatory to obtain a final grade equal to or higher than 5 points (out of 10).

ASSESSMENT ACTIVITIES

1. Multiple-choice exam: A multiple-choice exam will be held at the end of the scheduled teaching period (see course calendar date), which students must complete individually. This test will include multiple-choice

questions proposed by the different professionals who have been part of the course teaching staff, aimed at assessing mastery of the concepts and knowledge covered in class. This assessment accounts for 30% of the final course grade and is mandatory.

2. Presentation and defense of a case study: Students must individually prepare a case study, which will be submitted via the virtual campus to the course coordinator. Each student must present and orally defend the submitted case study. The assessment of this activity will take into account the design and formulation of the case, the oral presentation, the defense in response to questions, and participation in the discussion of other case studies. This activity accounts for 60% of the final grade (40% preparation and presentation, 20% defense) and is mandatory.

3. Career development plan: Students must prepare, according to their interests, a short-, medium- and long-term career development plan, following the framework provided in the relevant sessions. This activity accounts for 10% of the final course grade.

4. Attendance:

Given that attendance to sessions is mandatory, the following correction will be applied to determine the final grade:

- 100-75% attendance: the final grade will correspond to 100% of the score obtained in the assessment activities.
- $\leq 75\%$ - $\geq 50\%$ attendance: the final grade will correspond to 75% of the score obtained in the assessment activities.
- $< 50\%$ attendance: the final grade will correspond to 50% of the score obtained in the assessment activities.

Retake exam: A resit examination may be offered for students who have not achieved the minimum passing grade (5 out of 10).

Exam review: Exam reviews will be arranged by appointment on dates proposed by the teaching staff.

Single assessment: This subject/module does not foresee a single assessment system.

Not assessable: A student will be considered "not assessable" when they have not been able to provide the mandatory assessment evidence required for evaluation.

Bibliography

- McGowan-Jordan, J., Simons, A., & Schmid, M. (Eds.). (2020). *ISCN 2020: An International System for Human Cytogenomic Nomenclature*. Karger. <https://iscn.karger.com>
- Gardner, R. J. M., & Sutherland, G. R. (2012). *Chromosome Abnormalities and Genetic Counseling* (6th ed.). Oxford University Press.
- Rooney, D. E., & Czepulkowski, B. H. (Eds.). (2001). *Human Cytogenetics: Constitutional Analysis: A Practical Approach* (Vol. 240). Oxford University Press.
- Trounson, A., & Gardner, D. K. (Eds.). (2014). *Handbook of In Vitro Fertilization* (4th ed.). CRC Press.
- Falcone, T., & Hurd, W. (Eds.). (2017). *Clinical Reproductive Medicine and Surgery* (3rd ed.). Elsevier.
- European Society of Human Reproduction and Embryology (ESHRE). (2024). *Guidelines, consensus documents and recommendations*. <https://www.eshre.eu/Guidelines-and-Legal>
- De Geyter, C. (Ed.). (2020). *ESHRE PGT Consortium Data and Reports*. <https://www.eshre.eu/Data-collection-and-research/Consortia/PGD-Consortium/PGD-Consortium-Publicat>

The specialists in each session may propose additional bibliographic references.

Software

There is no specific software for this course

Course groups and languages

The information provided is provisional until November 30. After this date, you will be able to consult the language of each group through this [link](#). To access the information, you will need to enter the course CODE

Type of teaching	Group	Language	Semester	Shift
(TEm) Theory (master)	1	Catalan	second semester	afternoon
(SEMm) Seminars (master)	1	Catalan	second semester	afternoon