

Cytogenetics

Code: 101888
ECTS Credits: 6

2024/2025

Degree	Type	Year
2501230 Biomedical Sciences	OT	4

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Teachers

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Teaching groups languages

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

Prerequisites

Knowledge required to follow up on the subject:

1. To understand the basics of the subjects "Cell Biology" and "Genetics".
2. To understand specific aspects of these subjects: Mendelian principles, chromosome theory of inheritance, the flow of genetic information, the cell cycle principles and the mechanisms of cell division.
3. To read correctly in English.
4. To use at the user level, basic computer tools (Internet, PowerPoint, and Word Processor).

Objectives and Contextualisation

Cytogenetics is a hybrid discipline that draws on concepts of Cell Biology and Genetics. The convergence of issues from these areas has contributed to the development of a modern and dynamic science that has as a main objective the study of the chromosome.

The progress of this discipline has been characterized by the combination of conventional and modern techniques, as well as a continuous exchange between the development of new methods and the formulation of new hypotheses. This has significantly improved the understanding of the chromosome, providing a dynamic conception of this cell structure and developing to the limits the structure-function binomial.

In recent years, the consolidation of Cytogenetics has resulted in an alive discipline, approaching the borders of other disciplines with significant repercussions and applications in human health, agriculture, and evolution.

In this context, the objectives of the subject are:

1. To offer a comprehensive view into the structure and behavior of chromosomes to guarantee the preservation of genetic information, its transmission from parents to children, and gene expression.
2. To study chromosome variations, from the mechanisms that originate them to the genetic consequences for the offspring.
3. To perform a comprehensive analysis of the cytogenetics applications in human health, agricultural genomics, and speciation studies.

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.
- Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.
- Students must be capable of applying their knowledge to their work or vocation in a professional way and they should have building arguments and problem resolution skills within their area of study.
- Students must be capable of collecting and interpreting relevant data (usually within their area of study) in order to make statements that reflect social, scientific or ethical relevant issues.
- Students must be capable of communicating information, ideas, problems and solutions to both specialised and non-specialised audiences.
- Students must develop the necessary learning skills to undertake further training with a high degree of autonomy.
- Students must have and understand knowledge of an area of study built on the basis of general secondary education, and while it relies on some advanced textbooks it also includes some aspects coming from the forefront of its field of study.
- Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
- Work as part of a group with members of other professions, understanding their viewpoint and establishing a constructive collaboration.

Learning Outcomes

1. Apply the basic techniques commonly used in the cytogenetics laboratory.
2. Contrast the techniques and methods that allow genetic diagnosis.
3. Correctly use the terminology of genetics and its text and reference books
4. Describe the genetic techniques for the study and prevention of sterility and infertility.
5. Identify chromosome variants and anomalies, understand the mechanisms that originate them and determine the risk of their transmission to descendants.
6. Interpret scientific publications and solve problems and typical cases in the area of cytogenetics.
7. Interpret the forms of specialised chromosomes from the development of the structure-function binomial.
8. Make changes to methods and processes in the area of knowledge in order to provide innovative responses to society's needs and demands.

9. Recognise genetic anomalies in spermatogenesis and oogenesis related to a phenotype of sterility.
10. Recognise the anomalies of human chromosomes and assess their consequences.
11. Select the applications of cytogenetics in the field of the evolution of species, in the improvement of human health and in the genetic improvement of plants.
12. Students must be capable of applying their knowledge to their work or vocation in a professional way and they should have building arguments and problem resolution skills within their area of study.
13. Students must be capable of collecting and interpreting relevant data (usually within their area of study) in order to make statements that reflect social, scientific or ethical relevant issues.
14. Students must be capable of communicating information, ideas, problems and solutions to both specialised and non-specialised audiences.
15. Students must develop the necessary learning skills to undertake further training with a high degree of autonomy.
16. Students must have and understand knowledge of an area of study built on the basis of general secondary education, and while it relies on some advanced textbooks it also includes some aspects coming from the forefront of its field of study.
17. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
18. Understand and describe the structure, morphology and dynamics of eukaryote chromosomes in the different stages of the cell cycle.
19. Work as part of a group with members of other professions, understanding their viewpoint and establishing a constructive collaboration.

Content

PART I: ORGANIZATION OF HEREDITARY MATERIAL IN EUCARYOTES

Chapter 1. Overview

Chapter 2. The eukaryotic chromosome

PART II: CHROMOSOMES AND CELL DIVISION

Chapter 3. Mitotic cell division

Chapter 4. Meiotic cell division

PART III: SPECIALIZED CHROMOSOMES

Chapter 5. Adaptational forms of normal chromosomes

Chapter 6. Permanently specialized chromosomes

PART IV: TECHNIQUES FOR CHROMOSOME IDENTIFICATION AND ANALYSIS

Chapter 7. The basics of the cytogenetic analysis

Chapter 8. Chromosome identification techniques

PART V: GENETIC AND EPIGENETIC ANOMALIES

Chapter 9. Alterations of the karyotype

Chapter 10. Chromosome structural anomalies

Chapter 11. Chromosome numerical anomalies

Chapter 12. Epigenetic anomalies

PART VI: CYTOGENETICS APPLICATIONS

Chapter 13. Applications in speciation studies

Chapter 14. Applications in human health

Chapter 15. Applications in plant breeding

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Classroom practical classes (solved problems exposition)	5	0.2	17, 18, 5, 7, 6, 8, 16, 15, 14, 12, 13, 10, 9, 11, 19, 3
Laboratory practices	12	0.48	1, 2, 4, 6, 16, 15, 14, 13, 10, 19, 3
Theoretical classes	35	1.4	17, 18, 4, 5, 7, 8, 16, 15, 14, 10, 9, 11, 3
Type: Autonomous			
Individual study	60	2.4	18, 2, 4, 5, 7, 8, 16, 15, 14, 12, 10, 9, 11, 3
Solve problems	34	1.36	17, 18, 4, 5, 7, 6, 8, 15, 14, 12, 13, 10, 9, 11, 19, 3

Theoretical classes: The content of the theoretical program will be given by the teacher as master classes, with audio-visual support and encouraging the active participation of students through reciprocal questions. This teaching methodology will be applied in 50-minute sessions. The tables, figures and graphics used in class will be available in *pdf format on the Virtual Campus. Students may also consult on this platform the videos, animations and websites used in class. The study of the theory program implies that students have to regularly consult the books and review articles selected by the teacher (see Bibliography section). The articles will be available on the Virtual Campus in *pdf format.

Problem-solving classes: This learning methodology has the following objectives:

- To initiate the student in the resolution of representative experiments that clearly illustrate new advances in cytogenetics.
- To consolidate the concepts developed in theoretical 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.

There are 5 sessions of 50 minutes of duration programmed during the course. Within each group, students will be organized into groups of four people. The methodology applied to the classroom will consist of the following phases:

- Students will have a list of exercises (available on the Virtual Campus) that will be resolved in a non-face-to-face way by the established groups. For each of the scheduled sessions, students must work on 4 exercises and prepare an answer dossier.
- At the beginning of each session, each group will deliver to the teacher the answer dossier (one delivery per group). All exercises will be discussed and corrected in the classroom requiring the active participation of the students. Specifically, the teacher will ask a member of the different workgroups to present the resolution of an exercise to the rest of the students. The resolution will be evaluated by the

teacher and the qualification obtained will apply to all the members of the workgroup to which the student belongs.

- The answer dossier given by each workgroup will be evaluated as follows: at the end of each session, the teacher will choose a problem, which will be the same for all the groups, and these will be corrected and evaluated by the teacher.

The qualification obtained in Problem-solving activities will be the same for all the members of the group and will contribute to the final qualification of the subject.

Laboratory practices: Practices will be carried out in groups of two people. The students will have a guideline document (Virtual Campus of the subject) to address practical sessions. To facilitate the understanding of the contents and good development of the classes, it is advisable that the student read the practice guideline before each session. During the elaboration of the practices, students will have to solve face-to-face exercises facilitated by the teacher.

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	Weighting	Hours	ECTS	Learning Outcomes
Delivery and presentation of solved problems	15	0	0	17, 18, 2, 4, 5, 7, 6, 8, 16, 15, 14, 12, 13, 10, 9, 11, 19, 3
Delivery of exercises of the laboratory practices	15	0	0	1, 18, 2, 4, 6, 15, 14, 12, 13, 10, 19, 3
Written examination I (individual assessment)	35	2	0.08	18, 2, 4, 5, 7, 6, 16, 15, 14, 10, 9, 11, 3
Written examination II (individual assessment)	35	2	0.08	18, 2, 4, 5, 7, 6, 16, 15, 14, 10, 9, 11, 3

To pass the subject it is mandatory to obtain a final grade equal to or greater than 5 points out of 10 based on the contributions of the different evaluation activities. Students who perform fewer than 50% of the evaluation activities will be considered as not evaluated.

CONTINUOUS EVALUATION

1. Written exam (individual evaluation):

During the semester, two written tests (see course program) will be carried out on the theoretical contents of the subject. The objective is to evaluate the mastery of the concepts and the knowledge exposed in class, verifying the ability to apply and relate them.

Each test will weigh 35% on the final qualification of the subject. Students must obtain a minimum score of 4 points of the mean of the two written exams. To participate in the retake process students should have been previously evaluated in a set of activities equaling at least two-thirds of the final score of the course.

2. Problem-solving (group evaluation)

The qualification of this part will be obtained by the mean of the qualification obtained by each group of students throughout the course (dossier resolutions) and adding or subtracting up to 1 point out of 10 based on the contributions during the oral resolutions of the problems during the class. The teacher will ensure that each group has made at least one presentation. A problem not delivered or not solved in class will be scored with a zero in the calculation of the average grade of the group.

The assessment of the problems will be performed taking into account the correction in the response, the approach, and the interpretation of the results. The final grade will be shared by all the members of each group and will weigh 15% in the final mark of the subject.

3. Laboratory practices (group evaluation)

The laboratory practical mark will be obtained by the mean of the marks achieved in the exercises performed during the laboratory sessions. The assessment of the exercises will be performed by taking into account the correction in the response, the approach, and the interpretation of the results. The practical mark will be shared by all the members of each group and will be equivalent to 15% of the final mark.

Taking into account that practical sessions attendance is mandatory, an unjustified absence implies a penalty in the mark applying the following criteria:

- To miss one day implies a reduction of 30% in the laboratory practice mark.
- To miss two or more days implies a zero on the practice mark.

Students who can not attend their group session due to a justified cause are exempt from this penalty. The justified cause is understood to be health problems (the corresponding medical certificate must be brought to the coordinator of the practices) or serious personal problems. In this case, the practice will be recovered whenever possible.

UNIQUE EVALUATION

The unique evaluation consists of a synthesis written test in which the contents of the entire theoretical program of the subject will be assessed. This test will weigh 70% on the final qualification of the subject. This test will be held on the same date fixed in the calendar for the second exam of the continuous evaluation, and the same recovery system will be applied as for the continuous assessment. In order to use the qualification obtained in this synthesis test to calculate the final mark of the subject, students must obtain a minimum score of 4 points in this exam.

The evaluation of problem-solving and laboratory practices will be carried out in the same way as described in the continuous evaluation, and will also have the same weight in the final mark.

Bibliography

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Free electronic resource:

<http://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View..ShowTOC&rid=mboc4.TOC&depth=2>

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Software

No specific software is used.

Language list

Name	Group	Language	Semester	Turn
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(PAUL) Classroom practices	141	Catalan	second semester	morning-mixed
(PLAB) Practical laboratories	141	Catalan	second semester	afternoon
(TE) Theory	14	Catalan	second semester	morning-mixed