UAB
Universitat Autònoma de Barcelona

Cytogenetics

Code: 101964
ECTS Credits: 6

2024/2025

Degree	Type	Year
2500890 Genetics	OB	2

Contact

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Teachers

Joan Blanco Rodriguez

Teaching groups languages

You can view this information at the end of this document.

Prerequisites

Knowledge required:

1. To understand the basics of the first-year subjects "Cell Biology and Histology" and "Genetics".

2. To understand specific aspects of these subjects: Mendelian principles, chromosomal theory of inheritance, the flow of genetic information, the cell cycle, and mechanisms of cell division.

3. To have a medium English level.

Objectives and Contextualisation

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

The progress of this discipline has been characterized by the application of conventional and modern techniques, as well as a continuous dialogue 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 with significant social repercussions.

In this context, the specific objectives of the subject are:

1. To offer a comprehensive view of the structure and behavior of chromosomes to understand how chromosomes guarantee the preservation of genetic information, its transmission, and expression.

2. To study chromosome variations, from their mechanisms of origin to their genetic consequences.

Moreover, practical training in Integrated Laboratory III will complement the knowledge acquired in this subject.

Competences

- Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
- Apply knowledge of theory to practice.
- Apply scientific method to problem solving.
- Assume ethical commitment
- Be able to analyse and synthesise.
- Be able to communicate effectively, orally and in writing.
- 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.
- 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.
- Reason critically.
- Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
- Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
- Understand and describe the structure, morphology and dynamics of the eukaryotic chromosome during the cell cycle and meiosis.
- Use and manage bibliographic information or computer or Internet resources in the field of study, in one's own languages and in English.

Learning Outcomes

1. Act with ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
2. Apply knowledge of theory to practice.
3. Apply scientific method to problem solving.
4. Apply the basic common techniques used in the cytogenetics laboratory.
5. Assume ethical commitment
6. Be able to analyse and synthesise.
7. Be able to communicate effectively, orally and in writing.
8. Determine the mechanisms that cause chromosomal anomalies.
9. Develop self-directed learning.
10. Expose the new ethical dilemmas created by genetic progress.
11. Identify chromosomal variants and anomalies.
12. Identify the structure, morphology and dynamics of the eukaryotic chromosome in the different stages of the cell cycle.
13. Interpret the forms of specialised chromosomes from the structure-function binomial.
14. List and describe the applications of cytogenetics to the evolution of species, the improvement of human health and the genetic improvement of plants.
15. Quantify the risk of transmitting chromosomal anomalies to descendants.
16. Reason critically.
17. Resolve problems and example cases in the field of cytogenetics.

18. Take account of social, economic and environmental impacts when operating within one's own area of knowledge.
19. Take sex- or gender-based inequalities into consideration when operating within one's own area of knowledge.
20. Use and manage bibliographic information or computer or Internet resources in the field of study, in one's own languages and in English.

Content

PART I: ORGANIZATION OF HEREDITARY MATERIAL IN EUCARYOTES

Chapter 1. General 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 chromosomes

Chapter 6. Permanently specialized chromosomes

PART IV: TECHNIQUES FOR CHROMOSOME IDENTIFICATION AND CHROMOSOME ANALYSIS

Chapter 7. Basic principles of the cytogenetic laboratory techniques

Chapter 8. Chromosomal identification techniques

PART V: GENETIC AND EPIGENETIC ANOMALIES

Chapter 9. Alterations of the karyotype

Chapter 10. Structural chromosome anomalies

Chapter 11. Numerical chromosome anomalies

Chapter 12. Epigenetic anomalies

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Problem solving	7	0.28	2, 3, 4, 6, 7, 8, 11, 12, 13, 14, 15, 16, 17, 20
Seminar	3	0.12	4, 6, 7, 8, 11, 12, 13, 14, 15, 16, 17, 20
Theory	32	1.28	2, 4, 5, 8, 9, 11, 12, 13, 14, 15, 16, 17
Type: Autonomous			

Problem solving	30	1.2	2, 3, 4, 6, 7, 8, 10, 11, 12, 13, 14, 15, 17, 20
Seminar: oral presentation	15	0.6	4, 6, 7, 8, 10, 11, 12, 13, 14, 15, 16, 17, 20
Seminar: written presentation	15	0.6	4, 6, 7, 8, 10, 11, 12, 13, 14, 15, 16, 17, 20
Study	43	1.72	2, 4, 6, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 20

Theory classes

The content of the syllabus will be explained by the teache through lectures, supported by appropriate audiovisual materials, and encouraging active student participation through reciprocal questioning. This teaching methodology will be applied in 32 sessions of 50 minutes each. The tables, figures, and graphs used in class will be available in PDF format on the Moodle platform. Students will also be able to access videos, animations, and links to websites related to the subject on the Moodle platform. Following the program's theoretical content involves regular consultation of books and selected review articles by the professor to consolidate and clarify the topics covered in class (see Bibliography section). The articles will be available in PDF format on the Moodle platform.

Problem-solving Classes

The main objectives of this learning methodology are:

- Initiate students in solving representative experiments that illustrate recent advances in cytogenetics.
- Consolidate the concepts and knowledge covered in theory classes and evaluate their implications.
- Initiate students in the scientific method, focusing on learning objectives related to reasoning, critical judgment, and communication skills.

In these sessions, students will be divided into two groups. Students should check which group they belong to and attend the corresponding classes assigned to their group. Each group will have 7 sessions of 50 minutes each throughout the course. Within each group, students will organize themselves into workgroups of four. The classroom methodology will consist of the following phases:

- Students will receive a list of problems (available on the Moodle platform) that they will solve in their established groups remotely.
- For each scheduled session, students will work on 3 to 4 problems and prepare a response dossier.
- Within the indicated deadline on the Virtual Campus, and always before the face-to-face problem-solving class, each workgroup will submit their response dossier through the Moodle platform (one submission per group). These responses will then be discussed and corrected in class, with active participation from students. Specifically, the professor will randomly select a member from different workgroups to present the solution to a problem and explain it to the rest of the students. The problem-solving presentation will be evaluated by the professor, and the grade obtained will be applied to all members of the respective workgroup.
- The problem dossiers submitted by each group of students will be evaluated as follows: at the end of each session, the professor will choose a problem that will be the same for all groups, and they will be corrected and graded by the professor.
- The grade obtained will be the same for all members of the group and will contribute to the final grade of the subject.

Seminars

Among the competencies provided by the subject, taking into account the characteristics of the degree program and the students it is aimed at, it has been deemed appropriate to incorporate a competency related to the acquisition of the necessary skills for understanding and interpreting scientific publications. This is the fundamental objective of conducting seminars. In this regard, this activity will consist of preparing an oral

presentation and a written summary of a research article. Additionally, the seminars will aim to expand and consolidate different aspects of the theory and practical topics (Integrated Laboratory III) to deepen understanding through active student participation.

For the development of the seminars, students will organize themselves in the same way as for problem-solving classes. Each group of students will have 3 sessions of 50 minutes each throughout the course. The classroom methodology will consist of the following phases:

- At the beginning of the course, theprofessor will assign a scientific article to each workgroup.
- Based on the selected publication, each group will prepare a written work and an oral presentation. The presentation should be delivered within a maximum of 10 minutes, using audiovisual resources that the group considers appropriate.
- All members of the workgroup will give the public presentation, sharing the time equally. At the end of each presentation, the professor will ask two workgroups to each formulate one question.
- The written work will be submitted in PDF format on the day of the oral presentation and must adhere to the following format: maximum length of two pages; margins of 2.5 cm; font type Tahoma; font size 10; spacing of 6pt; single line spacing.
- The written work should answer the following questions regarding the content of the article: 1) What has been done? 2) Why has it been done? 3) How has it been done? 4) What are the most important results and interpretations? and 5) What is the conclusion?
- Workgroups that wish to do so may present and defend their work in English.

The grade obtained will be the same for all members of the workgroup and will contribute to the final grade of the subject.

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

Continous Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Problem solving	20	0.5	0.02	1, 2, 3, 4, 6, 7, 8, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20
Seminar oral and written presentation	10	0.5	0.02	1, 4, 6, 7, 8, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20
Written exam I	35	2	0.08	2, 4, 5, 6, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 20
Written exam II	35	2	0.08	2, 4, 5, 6, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 20

To pass the subject student must obtain a final mark equal to or greater than 5 points out of 10 based on the contributions of the different activities performed during the course. Moreover, students must obtain a minimum score of 4 points of the mean of the two written exams.

Students who do not reach the minimum qualification are eligible for the retake process, which will consist of a written exam (equivalent to those established by the partial tests). 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. Otherwise, students will obtain a qualification of non-evaluable.

Assessment activities:

- Written Exam (Individual Evaluation): Throughout the semester, two written tests (see course schedule) will be conducted on the theoretical content of the subject, which students will have to answer individually. The tests will consist of multiple-choice questions designed to assess the mastery of concepts and knowledge covered in class, as well as the ability to apply and relate them correctly. Each test (Written Exam I and II) will account for 35% of the final grade in the subject.
- Problem-solving (group assessment): The qualification of this part will be obtained by the arithmetic mean of the sum of the qualification obtained by each group of students throughout the course (dossier and oral resolutions). A problem not delivered or not resolved in class will be scored with a zero in the calculation of the average qualification of the group. The final qualification will be shared by all members of each group and will have a weight of 20% in the final mark of the subject.
- Seminars (group assessment): The qualification of this part will be obtained from the arithmetic mean of the oral and written presentations. This qualification will be modulated based on the quality of the questions/comments made by the students during the presentations of other groups. Besides, the use of English as a vehicle of communication will be assessed positively. The final mark will be shared by all the members of each group and will have a weight of 10%. This learning methodology will only contribute to the final mark whenthe students perform the two presentations (oral and written), otherwise, they will score with a zero.

Single evaluation

The students will be evaluated on the theoretical content of the subject through a single written exam (individual assessment), which will account for 70% of the final grade. The remaining 30% of the grade will be based on problem-solving and seminar participation, which will be assessed continuously throughout the course (see previous section).

This exam will be of the same format as the two exams scheduled for the continuous assessment of the theoretical content. It will be held on the same date specified in the calendar for Exam II, and the same recovery system as the continuous assessment will be applied.

Students who choose this option must inform the teacher before the first working day of October.

Bibliography

Alberts B, Johnson A, Lewis J, Raff M, Roberts K and Walter P (2022)* Molecular Biology of the Cell, 7th Edition. Garland Publishing, New York.

Free online book resource:

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

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Bickmore W and Craig J (1997) Chromosome bands: Patterns in the genome. Springer-Verlag Berlin Heidelberg, New York.

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Lodish H, Scott MP, Matsudaira P, Darnell J, Zipursky L, Kaiser CA, Berk A and Krieger M (2016) Molecular Cell Biology Eighth. WH Freeman Publishers, New York.

Free online book resource:

http://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View..ShowTOC&rid=mcb.TOC

Lynch M (2007) The Origins of Genome Architecture. Sinauer Associates Inc.

Rooney DE (2002) Human Cytogenetics: Constitutional Analysis. 3rd Edition. Oxford University Press. Oxford.

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Warshawsky D and Landolph JR. (2006). Molecular Carcinogenesis and the Molecular Biology.

Software

To consult the teaching material provided by the teaching staff, students must have programs that allow opening documents in pdf format.

Language list

Name	Group	Language	Semester	Turn
(PAUL) Classroom practices	621	Catalan	first semester	morning-mixed
(PAUL) Classroom practices	622	Catalan	first semester	morning-mixed
(TE) Theory	62	Catalan	first semester	afternoon