Universitat Autònoma de Barcelona

2022/2023

Cytogenetics

Code: 101964
ECTS Credits: 6

Degree	Type	Year	Semester
2500890 Genetics	OB	2	1

Contact

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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

Other comments on languages

L'activitat Seminari es desenvoluparà preferentment en anglès

Teachers

Joan Blanco Rodriguez

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.

4. To use basic informatics tools.

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

SYLLABUS 2022-2023

PART I: ORGANIZATION OF HEREDITARY MATERIAL IN HIGHER 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

Methodology

Theory classes

The content of the syllabus will be explained by the teacher in the form of lectures, with the appropriate audio-visual support and encouraging the active participation of students through reciprocal questions. This teaching methodology will be applied in 32 sessions of 50 minutes.

The tables, figures, and graphics used in class will be available in *pdf format on Moodle. Students may also consult on this platform the videos, animations, and websites used in class.

To consolidate the content explained in class, students will be requested to regularly consult books and review articles selected by the teacher. The articles will be available on Moodle in *pdf format.

Problem-solving classes

This learning methodology has the following objectives:

- To initiate the student in the resolution of representative experiments that illustrate new advances in cytogenetics.
- 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.

Students will be divided into two groups. Students must consult which group they belong to and attend classes in the assigned group. There are 7 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 dossier of problems (available on Moodle) 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 3-4 problems and prepare an answer dossier.
- Within the period indicated on the Virtual Campus, and always before the face-to-face class of problems, each group will deliver the answer dossier (only one delivery per group) using Moodle. All problems 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 a problem to the rest of the students. The resolution of the problem will be evaluated by the teacher and the qualification obtained will be applicable to all the members of the workgroup.
- 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 the Problem-solving activity will be the same for all the members of the group and will contribute to the final qualification of the subject.

Seminar classes

Among the competencies provided by the subject, and taking into account the characteristics of the Degree and the students to whom it is addressed, it has been considered appropriate to incorporate a competence related to the acquisition of the skills necessary for the understanding and interpretation of scientific publications. This is the basic objective of seminars. In this sense, this activity consists of a discussion by means of an oral and a written presentation of a research paper. The seminars will extend and consolidate different aspects treated in the theory and practice sessions (Integrated Laboratory III) through the active participation of the students.

For the preparation of the Seminars, the students will be organized as well as for the Problem-solving classes. Each group of students will complete 3 sessions of 50 minutes during the course. The methodology applied in the classroom will consist of the following phases:

- At the beginning of the course, the teacher will assign a scientific paper to each workgroup and each group has to prepare an oral and a written presentation.
- The oral presentation will last less than 10 minutes using the audio-visual support selected by the group.
- The written work will be delivered in *pdf format the day of the oral presentation and must be written using the following format: Maximum length of two pages; margins 2.5 cm; Tahoma font; size 10; 6pt spacing; simple spacing.
- The written work must answer the following questions in relation to the content of the article: 1) What is the problem?, 2) Why it is important?, 3) How do the authors study the problem?, 4) What are the most important results and interpretations? and 5) What is the conclusion?

Working groups can give the presentation in English.

The qualification obtained will be the same for all the members of the group and will contribute to the final qualification 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.

Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Problem solving	7	0.28	3, 2, 4, 8, 12, 11, 13, 14, 15, 16, 17, 7, 6, 20
Seminar	3	0.12	4, 8, 12, 11, 13, 14, 15, 16, 17, 7, 6, 20
Theory	32	1.28	2, 4, 5, 9, 8, 12, 11, 13, 14, 15, 16, 17
Type: Autonomous			
Problem solving	30	1.2	3, 2, 4, 8, 10, 12, 11, 13, 14, 15, 17, 7, 6, 20
Seminar: oral presentation	15	0.6	4, 8, 10, 12, 11, 13, 14, 15, 16, 17, 7, 6, 20
Seminar: written presentation	15	0.6	4, 8, 10, 12, 11, 13, 14, 15, 16, 17, 7, 6, 20
Study	43	1.72	2, 4, 9, 8, 10, 12, 11, 13, 14, 15, 16, 17, 6, 20

Assessment

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 assessment): During the semester two written exams will be carried out to assess the knowledge of the subject (see syllabus). Each test will have a weight of 35% on the final mark.
- 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.

Assessment Activities

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

Bibliography

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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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Software

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