Universitat Autònoma de Barcelona

2019/2020

Cytogenetics

Code: 101964
ECTS Credits: 6

Degree	Type	Year	Semester
2500890 Genetics	OB	2	1

Contact

Name: Joan Blanco Rodríguez
Email: Joan.Blanco@uab.cat

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

Prerequisites

Knowledge required:

1. Understand the basics of the subjects: "Cell Biology and Histology" and "Genetics".

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

3. Read correctly in English.

4. 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 improve 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 to 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 chromosomes variations, from the mechanisms that originate them to the genetic consequences for the offspring.

Moreover, practical training in Integrated Laboratory III will complement the theoretical knowledge acquired during the course.

Competences

- 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.
- 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 ones own languages and in English.

Learning Outcomes

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

Content

PART I: ORGANIZATION OF HEREDITARY MATERIAL IN HIGHER EUCARYOTES

Chapter 1. General introduction

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. Generalities of the cytogenetic analysis protocols

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

Methodology

Theory classes

The content of the theory program will be explained by the teacher in the form of master classes, 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 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 regularly consult the books and review articles selected by the teacher in order to consolidate the contents explained in class (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 inititiate the student in the resolution of representative experiments that clearly 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. The student must consult which group they belong to and attend the classes in their assigned group. There are 7 sessions of 50 minutes of duration programmed during the course. Within each group, students will be organized in groups of four people. The methodology applied to the classroom will consist on the following phases:

- Students will have a list of problems (available at 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 problems and prepare an answer dossier.
- At the beginning of each session, each group will deliver the teacher the answer dossier (one delivery per group). 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 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.

Seminar classes

Among the competences provided by Cytogenetics, and taking into account the characteristics of the Degree and the students to which it is addressed, we believe it is appropriate to incorporate one activity related to the provision of tools for the understanding and interpretation of scientific publications. This is the basic objective of seminars. This activity consist 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 during the course 3 sessions of 50 minutes. The methodology applied in the classroom will consist on the following phases:

- At the beginning of the course the teacher will assign a scientific paper to eachworkgroup and each group have to prepare a written and an oral presentation.
- The oral presentation will consist of 6 slides that will be presented to the rest of the students with a maximum time of 7 minutes.
- The written work will be delivered in *pdf format the day of the oral presentation and will have to be adjusted to 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 did the authors study the problem?, 4) What are the most important results and interpretations? and 5) What is the conclusion?

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

Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Problem solving	7	0.28	2, 1, 3, 7, 11, 10, 12, 13, 14, 15, 16, 6, 5, 17
Seminar	3	0.12	3, 7, 11, 10, 12, 13, 14, 15, 16, 6, 5, 17
Theory	32	1.28	1, 3, 4, 8, 7, 11, 10, 12, 13, 14, 15, 16
Type: Autonomous			
Problem solving	30	1.2	2, 1, 3, 7, 9, 11, 10, 12, 13, 14, 16, 6, 5, 17
Seminar: oral presentation	15	0.6	3, 7, 9, 11, 10, 12, 13, 14, 15, 16, 6, 5, 17
Seminar: written presentation	15	0.6	3, 7, 9, 11, 10, 12, 13, 14, 15, 16, 6, 5, 17
Study	43	1.72	1, 3, 8, 7, 9, 11, 10, 12, 13, 14, 15, 16, 5, 17

Assessment

To pass the subject it will be essential to obtain a final qualification equal or greater than 5 points out of 10 based on the contributions of the different evaluation activities. In addition, 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, that will consist in 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 tests will be carried out on the theoretical contents of the subject (see programming). These tests will consist of a series of test-type questions that students must answer individually. Each test will have a weight of 35% on the final qualification of 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. The final qualification 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 when the 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	2, 1, 3, 7, 9, 11, 10, 12, 13, 14, 15, 16, 6, 5, 17
Seminar oral and written presentation	10	0.5	0.02	3, 7, 9, 11, 10, 12, 13, 14, 15, 16, 6, 5, 17
Written exam I	35	2	0.08	1, 3, 4, 8, 7, 9, 11, 10, 12, 13, 14, 15, 16, 5, 17
Written exam II	35	2	0.08	1, 3, 4, 8, 7, 9, 11, 10, 12, 13, 14, 15, 16, 5, 17

Bibliography

Alberts B, Johnson A, Lewis J, Raff M, Roberts K and Walter P (2008)* Molecular Biology of the Cell, 5th Edition. Garland Publishing, New York.

Andreeff M and Pinkel D (1999) Fluorescence in situ hybridization: Principles and clinical apllication. Wiley-Liss. New York.

Bickmore W (1999)* Chromosome Structural Analysis; A Practical Approach. Oxford University Press, Oxford.

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

Gardner RJM and Sutherland GR (2004)* Chromosome Abnormalities and Genetic Counseling, 3rd edition. Oxford University Press. Oxford.

Holmquist GP and Motara MA (1987) The magic of cytogenetic technology. In Cyogenetics. Obe G and Basler A Editors. Springer-Verlag, Berlin.

King M (1993)* Species evolution. The role of chromosome change. Cambridge University Press.

Lacadena JR (1996)* Citogenética. Editorial Complutense SA, Madrid.

Lodish H, Scott MP, Matsudaira P, Darnell J, Zipursky L, Kaiser CA, Berk A and Krieger M (2003)* Molecular Cell Biology. WH Freeman Publishers, New York.

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

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

Singh RJ (2002) Plant cytogenetics. CRC Press.

Solari AJ. (2004)* Genética Humana. Fundamentos y Aplicaciones en Medicina. 3ª edición. Médica Panamericana. Buenos Aires.

Sumner AT (2003)* Chromosomes: Organization and Function. Blackwell Publishing.

Sybenga J (1975)* General Cytogenetics. North-Holland Publishing Company. Amsterdam.

Sybenga J (1975)* Meiotic Configurations. Springer-Verlag Berlin Heidelberg. New York.

Tost J (2007) Epigenetics. Caister Academic Press.

Turner J (2007)* Meiosis. Chromosome research 15. Special issue (5). Springer.

Vogelstein B and Kinzler KW (2002) The Genetic Basis of Human Cancer. 2nd Edition. Graw-Hill Professional. New York.

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