Universitat Autònoma de Barcelona

2018/2019

New Developments in Cytogenetics and Biology of Reproduction

Code: 42941
ECTS Credits: 6

Degree	Type	Year	Semester
4313782 Cytogenetics and Reproductive Biology	OB	0	1

Contact

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Use of languages

Principal working language: catalan (cat)

Prerequisites

The same prerequisites for admission to the Master

Objectives and Contextualisation

This is a compulsory course that aims to introduce the latest concepts and methodologies related to the fields of cytogenetics and reproductive biology for all students taking the Masters in Cytogenetics and Reproductive Biology.

The specific goals of the course are:

1.-Understand the structure, regulation and orgaització of the mammalian genome.

2.-Understand the process of differentiation and interaction of mammalian gametes that leads to the formation of an embryo.

Skills

- Apply the scientific method and critical reasoning to problem solving.
- Communicate and justify conclusions clearly and unambiguously to both specialist and non-specialist audiences.
- Continue the learning process, to a large extent autonomously.
- Identify the cellular and molecular bases of human pathologies linked to chromosome anomalies.
- Integrate knowledge and use it to make judgements in complex situations, with incomplete information, while keeping in mind social and ethical responsibilities.
- Recognise the cellular and molecular bases of reproduction in mammals.
- Solve problems in new or little-known situations within broader (or multidisciplinary) contexts related to the field of study.
- Use acquired knowledge as a basis for originality in the application of ideas, often in a research context.
- Use and manage bibliography or ICT resources in the master's programme, in one's first language and in English.

Learning outcomes

1. Apply the scientific method and critical reasoning to problem solving.

2. Communicate and justify conclusions clearly and unambiguously to both specialist and non-specialist audiences.
3. Continue the learning process, to a large extent autonomously.
4. Describe the cellular and molecular processes of fertilisation and pre-implantation embryo development.
5. Describe the epigenetic regulation that conditions the function of centromeres and telomeres.
6. Describe the structure, dynamics and morphology of the eukaryote chromosome at any stage of the cell cycle and during meiosis.
7. Identify chromosome anomalies, understand the mechanisms that cause them and determine the risk of transmission to offspring.
8. Identify the cellular and molecular bases of human spermatogenesis and ovogenesis.
9. Integrate knowledge and use it to make judgements in complex situations, with incomplete information, while keeping in mind social and ethical responsibilities.
10. Recognise the fundamental role of immunology in human reproduction
11. Recognise the influence of chromatin fibre in gene expression.
12. Solve problems in new or little-known situations within broader (or multidisciplinary) contexts related to the field of study.
13. Use acquired knowledge as a basis for originality in the application of ideas, often in a research context.
14. Use and manage bibliography or ICT resources in the master's programme, in one's first language and in English.

Content

Unit 1: Advanced Complements in Cytogenetics. Organization of chromatin fiber: in silico and in vivo studies. Chromosome territories, nuclear architecture and gene regulation in higher eukaryotes. Epigenetic regulation of chromosome function. Origin and recurrence of human diseases caused by chromosomal abnormalities.

Unit 2: Advanced Complements in Reproductive Biology. Cellular and molecular aspects of male and female gametogenesis. Acquisition of the fertilizing capacity of sperm. Mechanisms of interaction between male and female gametes. Immunology of male and female reproductive tract.

Methodology

The contents of this course include lectures, taught by academics and / or professionals, which will encourage student participation.

Activities

Title	Hours	ECTS	Learning outcomes
Type: Directed			
Lectures	32	1.28	4, 6, 7, 8, 9, 12, 3, 10, 11, 14
Type: Autonomous			
Study	104	4.16	4, 6, 5, 7, 8, 9, 3, 10, 11, 14

Evaluation

The skills of this course will be evaluated in two sections:

1.-Written test (80% of grade): two multiple-choice test will evaluate the knowledge acquired by each student. The mark obtained in each test will represent 40% of the final mark. These tests will be performed at the end of the program contents.

2-Participation in class (20% of grade): An evaluation of student participation during lectures in the debates raised by teachers will be performed.

In case of obtaining less than a 5 in the final grade, a multiple-choice examn will be carried out.

Evaluation activities

Title	Weighting	Hours	ECTS	Learning outcomes
Participacition in the classroom	20%	10	0.4	1, 9, 12, 2, 13, 14
Written test	80%	4	0.16	1, 4, 6, 5, 7, 8, 9, 12, 3, 10, 11, 13, 14

Bibliography

River RM, Bennet LB. Epipgenetics in humans: an overview. Curr Opin Endocrinol Diabetes Obse. 2010. 17(6):493-9

Editorial i articles inclosos a Mol Hum Reprod. 2010. 16 (1):1-56

Chowdhury D, Choi YE, Brault ME. Charity begins at home: non-coding RNA functions in DNA repair. Nat Rev Mol Cell Biol. 2013. 14(3):181-9

Hoeijmakers JHJ. Genome maintenance mechanisms for preventing cancer. Nature. 2001. 411(6835):366-74

Girirajan S, Dennis MY, Baker C, Malig M, Coe BP, Campbell CD, Mark K, Vu TH, Alkan C, Cheng Z, Biesecker LG, Bernier R, Eichler EE. Refinement and Discovery of New Hotspots of Copy-Number Variation Associated with Autism Spectrum Disorder. Am J Hum Genet . 2013. 92, 221-237

Mefford EC, Eichler EE. Duplication hotspots, rare genomic disorders, and common disease. Current Op in Genetics & Development. 2009. 19:196-204

Miller TD, Adam MP, Aradhya S, Biesecker LG, Brothman AR et al. Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. Am J Hum Genet. 2010. 86: 749-764.

Rodriguez-Santiago B, Armengol Ll. Tecnologías de secuenciación de nueva generación en diagnóstico genético pre- y postnatal. Diagnóstico prenatal. 2012. 23 (2): 56-66

Cooper GM, Coe BP, Girirajan S, Rosenfeld JA, Vu TH et al. A copy number variation morbidity map of developmental delay. Nature Genetics. 2011. 43 (9): 838-846.

The Nucleus. Edited by Tom Misteli, *National Cancer Institute/National Institutes of Health;* David L. Spector, *Cold Spring Harbor Laboratory.* Cold Spring Harbor Laboratory Press

Biochimica et Biophysica Acta (BBA)-Gene Regulatory Mechanisms. 2012. 1819 (7): 631-846(July2012). Special issue: Chromatin in time and space

Boulcun-Filas E, Schimenti J. Genetics of meiosis and recombination in mica. Int Rev Cell Mol Biol. 2012. 298:179-227

Nagaoka SI, Hassold TJ, Hunt PA. Human aneuploidy: mechanisms and new insights into an age-old problem. Nat Rev Genet. 2012. 13 (7):493-504

Jessberger R. Age-related aneuploidy through cohesion exhaustion. EMBO Rep. 2012. 13 (6): 539-46