UAB
Universitat Autònoma de Barcelona

Integrated Cytogenetics Laboratory

2026/2027

Code: 42950
Credits: 9

Degree programme	Type	Course
Cytogenetics and Reproductive Biology	OP	1

Contact lecturer

Name : Alejandro Gella Concustell

Email : alex.gella@uab.cat

Teaching staff

Javier Rey Azpiri

Joaquim Martí Clua

Andreu Blanquer Jerez

Berta Nieves Vazquez Prat

Maria Angels Rigola Tor

Aurora Ruiz-Herrera Moreno

Ignasi Roig Navarro

Alejandro Gella Concustell

Group languages

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

Prerequisites

The same prerequisites for admission to the Master.

Objectives

The module "Integrated Cytogenetics Laboratory" is a compulsory practical module within the Cytogenetics specialty. Its main objective is to provide students with the basic tools required to apply the cytogenetic techniques used both in genetic diagnostic laboratories and in research laboratories.

The learning objectives of this module are as follows:

1. To acquire the ability to work under sterile conditions in order to establish cell cultures.

2. To identify different types of contamination in cell cultures.

3. To obtain histological sections and apply different staining techniques.

4. To learn how to use different types of microscopes: bright-field microscope, fluorescence microscope, and laser scanning confocal microscope.

5. To identify human chromosomes according to the G-banding pattern and prepare a karyotype, as well as to detect alterations in this banding pattern.

6. To detect proteins and DNA sequences using immunocytofluorescence and fluorescence in situ hybridization (FISH) techniques, respectively.

7. To identify genes located in a specific region of the genome and design a fluorochrome-labelled DNA probe for application in FISH techniques.

Learning outcomes

- CA07 (Autonomously plan the management of research and clinical laboratories in the field of cytogenetics, integrating advanced knowledge and applying quality, biosafety and efficiency criteria in the organisation of resources.) Autonomously plan the management of research and clinical laboratories in the field of cytogenetics, integrating advanced knowledge and applying quality, biosafety and efficiency criteria in the organisation of resources.
- CA08 (Autonomously design scientific experiments in the field of cytogenetics, applying advanced methodologies and criteria of rigor, safety and ethics in research.) Autonomously design scientific experiments in the field of cytogenetics, applying advanced methodologies and criteria of rigor, safety and ethics in research.
- KA12 (Identify the cytogenetic origin and cellular foundations of human pathologies.) Identify the cytogenetic origin and cellular foundations of human pathologies.
- SA08 (Apply cell culture and characterisation techniques in a cytogenetics laboratory.) Apply cell culture and characterisation techniques in a cytogenetics laboratory.
- SA09 (Apply the principles of quality, biosafety and traceability when carrying out activities, using resources and monitoring workflows in an integrated cytogenetics laboratory.) Apply the principles of quality, biosafety and traceability when carrying out activities, using resources and monitoring workflows in an integrated cytogenetics laboratory.
- SA10 (Critically interpret research data from an integrated cytogenetics laboratory, integrating advanced knowledge to formulate well-supported conclusions.) Critically interpret research data from an integrated cytogenetics laboratory, integrating advanced knowledge to formulate well-supported conclusions.
- SA11 (Solve clinical cases in the field of clinical cytogenetics, applying laboratory techniques and cytogenetic analysis procedures.) Solve clinical cases in the field of clinical cytogenetics, applying laboratory techniques and cytogenetic analysis procedures.
- SA12 (Demonstrate advanced communication skills, both oral and written, to communicate results obtained in a clinical or cytogenetics research laboratory.) Demonstrate advanced communication skills, both oral and written, to communicate results obtained in a clinical or cytogenetics research laboratory.

Contents

Topic 1. Cytological and Histological Techniques

Organ embedding and fixation. Microtomy. Immunofluorescence. Various staining techniques. Microscopic visualization and image digitization. Image processing using ImageJ.

Topic 2. Cytogenetics and Immunofluorescence Techniques

Cell line culture for the obtention of metaphase chromosomes. Cultures for the fluorescent immunodetection of proteins. Preparation of FISH probes from BACs and YACs. Fluorescence in situ hybridization in different types of samples. Image acquisition using a fluorescence microscope. Detection of contamination in cell cultures.

Topic 3. Confocal Microscopy

Fundamentals of fluorescence and confocal microscopy. Preparation of samples for fluorescence. Image acquisition using the confocal microscope. Processing of image series with ImageJ.

Topic 4. Karyotyping and Chromosomal Analysis Techniques

Identification of human chromosomes according to the G-banding pattern. Identification of chromosomal alterations. Cytogenetic nomenclature.

Learning activities and methodology

Title	Hours	ECTS	Learning outcomes
Problems and pratical cases preparation	10	0.4	
Confocal laser scanning microscopy	10	0.4	
Photographic composition preparation	5	0.2	
Resolution of practical cases or problems	12	0.48	
Cell culture, fluorescence in situ hybridization (FISH) and immunocytofluorescence	35	1.4	
Report preparation of the practices results	20	0.8	
Study	63	2.52	
Personalized tutoring	30	1.2	
Update in histological tecniques	20	0.8	
Chromosome identification: karyotype	5	0.2	
Preparation of the practices reports	15	0.6	

The module "Integrated Cytogenetics Laboratory" has a mainly practical focus and is structured into four topics.

Teaching will include laboratory practical sessions, while Topic 3, devoted to confocal microscopy, will be carried out at the UAB Microscopy Service.

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	Weight	Hours	ECTS	Learning outcomes
Karyotyping and Chromosomal Analysis Techniques	7%	0	0	CA07, CA08, KA12, SA09, SA10, SA11, SA12

Cytogenetics and Immunofluorescence Techniques	50%	0	0	CA07, CA08, KA12, SA08, SA09, SA10, SA11
Cytological and Histological Techniques	29%	0	0	CA07, CA08, KA12, SA08, SA09, SA10, SA11, SA12
Confocal Microscopy	14%	0	0	CA07, CA08, SA09, SA10

To pass the module, students must obtain a minimum overall mark of 5 out of 10.

Attendance at practical sessions is compulsory.

The final mark will be calculated as the weighted sum of the marks obtained in each assessment block. The weight of each block will be proportional to the time allocated to the scheduled sessions for carrying out these activities.

In all blocks, students' attitude and active participation will account for 10% of the corresponding mark.

The competences of this module will be assessed through the following activities:

1. Cytological and Histological Techniques

- Participation and performance in practical sessions: 10%
- Individual submission of a report and questionnaire: 45%
- Preparation of a photographic composition using Photoshop: 45%

2. Cytogenetics and Immunofluorescence Techniques

- Participation and performance in practical sessions: 10%
- Submission of a report including the results obtained through the application of these techniques: 90%

3. Confocal Microscopy

- Participation and performance in practical sessions: 10%
- Written examination: 90%

4. Karyotyping and Chromosomal Analysis Techniques

- Participation and performance in practical sessions: 10%
- Resolution of normal and altered karyotypes using the Human Karyolab program: 40%
- Resolution and submission of karyotypes with abnormalities, indicating the formula, the clinical features of the abnormality, and the risk of having affected offspring: 50%

Permitted Use of Artificial Intelligence

The use of artificial intelligence tools is permitted solely as auxiliary support for linguistic improvement, spelling and grammar correction, translation, formal organisation of information, and document presentation. Their use is not permitted to replace students' own work in the interpretation of results, the completion of questionnaires, the preparation of karyotypes, image analysis, or the full drafting of reports. Students will be responsible for the final content submitted, must be able to justify the results and conclusions included in their assessment activities and, when AI tools are used, this must be stated in the submitted document.

Irregularities in Assessment Activities

Any irregularity committed in an assessment activity -academic fraud, plagiarism, or improper use of artificial

intelligence, unless such use has been expressly authorised- that may lead to a significant change in the mark will result in that assessment activity being marked as 0. In addition, disciplinary proceedings may be initiated against students who commit any of these irregularities.

Resit assessment

Given that the module "Integrated Cytogenetics Laboratory" is eminently practical in nature, no ordinary resit assessments are planned. However, exceptionally, the teaching staff responsible for each practical block, with the approval of the module coordination, may schedule a resit activity.

In order to take part in a resit activity, students must have previously been assessed in a set of activities whose weight is equivalent to at least two thirds of the total module mark. Therefore, students will receive a "Not assessable" mark when the assessment activities completed account for less than 67% of the final mark.

"Not assessable" mark

Students will receive a "Not assessable" mark in the following cases:

- When their absence exceeds 20% of the scheduled sessions.
- When the assessment activities completed account for less than 67% of the final mark.

This module does not provide for the single assessment system.

Bibliography

Books

- Animal Cell Culture Methods. Methods in Cell Biology.J.P. Mather and D. Barnes Eds. Academic Press. 1998
- Cell and Tissue Culture: Laboratory procedures in biotechnology.A. Doyle and J.B. Griffiths Eds. JohnWiley & Sons Ltd. 1999
- Culture of animal cells. A manual of basic technique (6th ed.) RI Freshney. Wiley-Liss, 2010
- Cytogenetic and genome research. RH Martin. Karger,2002
- Chromosome Abnormalities and genetic counseling (3rd ed). RJ McKinlay & GR Sutherland, Oxford University Prees, 2004
- ISCN 2016. An International System for Human Cytogenomic Nomenclature (2016). McGowan-Jordan J, Simons A, Schmid M, editors. Karger. 2016
- Theory and Practice of Histological Techniques (sixth edition). John D. Bancroft, Churchill Livingstone. Elsevier. 2008

Webs

- 29 Mammals Project - http://www.broadinstitute.org/scientific-community/science/projects/mammals-models/mammalian-genome
- Cytogenetic Resources http://www.kumc.edu/gec/prof/cytogene.html
- Discover Life - http://www.discoverlife.org/mp/20m?tree=Life&flags=all
- Ensembl - http://www.ensembl.org/index.html
- GeneReviews - http://www.ncbi.nlm.nih.gov/sites/GeneTests/review?db=GeneTests
- Genetics Home Reference - http://ghr.nlm.nih.gov/ghr/page/Home
- Genome 10K Project - http://genome10k.soe.ucsc.edu/
- Molecular Expressions. https://micro.magnet.fsu.edu/
- Olympus. Microscopy Resource Center. https://www.olympus-lifescience.com/en/microscope-resource/

- Online Mendelian Inheritance in Man (OMIM) - http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM
- Orphanet - http://www.orpha.net/consor/cgi-bin/home.php?Lng=ES
- PubMed - http://www.kumc.edu/gec/prof/cytogene.html
- The National Center for Biotechnology Information - http://www.ncbi.nlm.nih.gov/
- TIMETREE - http://timetree.org/index.php
- UCSC Genome Bioinformatics Site - http://genome.ucsc.edu/
- University of Wisconsin - http://www.slh.wisc.edu/wps/wcm/connect/extranet/cytogenetics
- Zeiss Campus. http://zeiss-campus.magnet.fsu.edu/

The specific bibliography corresponding to the different contents of the module may be requested to the responsible teachers of each block.

Software

ISIS image acquisition software: (MetaSystems, Altlussheim, Germany).

Ikaros image acquisition software: (MetaSystems, Altlussheim, Germany).

ImageJ: Schneider CA, Rasband WS, Eliceiri KW. "NIH Image to ImageJ: 25 years of image analysis". Nature Methods 9, 671-675, 2012.

Karyolab: Gibbons NJ, Evans C, Griffin DK. " Learning to karyotype in the university environment: a computer-based virtual laboratory class (KaryoLab) designed to rationalize time for the tutor/researcher and to encourage more students to engage in cytogenetics". Cytogenet Genome Res 101:1-4 (2003). https://doi.org/10.1159/000073409

Microsoft office

Course groups and languages

The information provided is provisional until November 30. After this date, you will be able to consult the language of each group through this link. To access the information, you will need to enter the course CODE

Type of teaching	Group	Language	Semester	Shift
(PAULm) Classroom practices (master)	1	Catalan	second semester	morning-mixed
(PLABm) Practical laboratories (master)	1	Catalan/Spanish	second semester	morning-mixed