Universitat Autònoma de Barcelona

2023/2024

Integrated Cytogenetics Laboratory

Code: 42950
ECTS Credits: 9

Degree	Type	Year	Semester
4313782 Cytogenetics and Reproductive Biology	OT	0	2

Contact

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Teaching groups languages

You can check it through this link. To consult the language you will need to enter the CODE of the subject.
Please note that this information is provisional until 30 November 2023.

Teachers

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Prerequisites

The same prerequisites for admission to the Master.

Objectives and Contextualisation

The module "Laboratori integrat de citogenètica" is a compulsory practical module of the Citogenetics specialty. The main objective is to provide students with the basic tools to apply cytogenetic techniques used in genetic diagnostic laboratories and in research laboratories.

The training objectives of this module are:

1- Acquire the ability to work in sterile conditions to establish a cell culture and obtain cells at different mitotic stages.

2- Identify different types of cell culture contamination.

3- Obtain histological tissue sections and apply different stains.

4- Learn to use different types of microscopes: clear field, fluorescence and confocal laser scanning.

5- Identify human chromosomes according to the pattern of G-bands and make the karyotype. Detect the alterations of this pattern of bands.

6- Detect proteins and DNA sequences using immunocytofluorescence and fluorescence *in situ* hybridization techniques (FISH), respectively.

7- Identify genes affected in a region of the genome and produce a fluorochrome labeled DNA probe to apply FISH techniques.

Competences

- Apply knowledge of theory in both research and clinical care contexts.
- 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.
- Design and execute analysis protocols in the area of the master's degree.
- Design experiments, analyse data and interpret findings.
- Integrate knowledge and use it to make judgements in complex situations, with incomplete information, while keeping in mind social and ethical responsibilities.
- Show an ability to work in teams and interact with professionals from other specialist areas.
- 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.
- Use creative, organisational and analytic skills when taking decisions.

Learning Outcomes

1. Apply histology techniques in different tissues of the organism.
2. Apply immunocytofluorescence techniques in different cell types.
3. Apply knowledge of theory in both research and clinical care contexts.
4. Apply the scientific method and critical reasoning to problem solving.
5. Apply the techniques of molecular cytogenetics in different cell types.
6. Communicate and justify conclusions clearly and unambiguously to both specialist and non-specialist audiences.
7. Continue the learning process, to a large extent autonomously.
8. Demonstrate an ability to work in sterile conditions in the culture laboratory.
9. Design experiments, analyse data and interpret findings.
10. Incorporate and make use of the information obtained from the different online databases on genome sequencing and localisation.
11. Integrate knowledge and use it to make judgements in complex situations, with incomplete information, while keeping in mind social and ethical responsibilities.
12. Recognise the different uses of a confocal laser microscope.
13. Show an ability to work in teams and interact with professionals from other specialist areas.

14. Solve problems in new or little-known situations within broader (or multidisciplinary) contexts related to the field of study.
15. Use acquired knowledge as a basis for originality in the application of ideas, often in a research context.
16. Use and manage bibliography or ICT resources in the master's programme, in one's first language and in English.
17. Use creative, organisational and analytic skills when taking decisions.

Content

1. Update on histological techniques

Development of histological technique: inclusion and microtomy.

Staining of histological sections.

Immunofluorescence.

Flow cytometry.

Microscopic visualization and digital imaging.

Image processing using Photoshop.

2. Cell culture, fluorescent *in situ* hybridization (FISH) and immunofluorescence

Cultures for obtaining metaphase chromosomes.

Cultures for Fluorescent immunodetection of proteins.

Generation of FISH probes from BACs and YACs.

Fluorescent *in situ* hybridization in different types of samples .

Detection of contamination in cell cultures.

3. Confocal laser scanning microscopy

Fundamentals of Fluorescence and Confocal Microscopy.

Fluorescence sample preparation.

Image capturing using Confocal Microscope.

Image processing using Fiji.

4. Chromosome identification: karyotype

Identification of human chromosomes by G-banding pattern.

Chromosomal alterations.

Cytogenetic nomenclature.

Methodology

The module "Integrated Cytogenetics Laboratory" is basically practical, distributed in 4 blocks. The practices will be done in the laboratory and the Microscopy Service of the UAB.

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			
Cell culture, fluorescence in situ hybridization (FISH) and immunocytofluorescence	35	1.4	4, 3, 2, 5, 13, 8, 9, 14, 16
Chromosome identification: karyotype	5	0.2	3, 13, 11, 14, 16
Confocal laser scanning microscopy	10	0.4	3, 13, 9, 12
Update in histological tecniques	20	0.8	4, 3, 1, 13, 9
Type: Supervised			
Personalized tutoring	30	1.2	11, 14, 6, 7
Photographic composition preparation	5	0.2	14, 7, 16
Preparation of the practices reports	15	0.6	3, 13, 17, 11, 14, 6, 7, 15
Problems and pratical cases preparation	10	0.4	4, 3, 13, 9, 17, 11, 14, 6, 7, 15, 16
Type: Autonomous			
Report preparation of the practices results	20	0.8	13, 9, 17, 11, 6, 7, 15, 16
Resolution of practical cases or problems	12	0.48	4, 3, 13, 8, 9, 17, 11, 14, 6, 7, 15, 16
Study	63	2.52	17, 11, 14, 6, 7, 15, 16

Assessment

In order to pass the module, it will be necessary to obtain a global qualification equal to or greater than 5 out of a maximum of 10.

Attendance at practical sessions is mandatory. The students will obtain the "No Avaluable" qualification when the absence exceeds 20% of the programmed sessions.

The final qualification results from the weighted sum of the mark obtained in each evaluation block. The weight of each evaluation block is proportional to the time spent in the sessions programmed to perform these activities. In each block, attitude and active participation represent 10% of the qualification.

The competences of this module will be evaluated as follows:

1. Update in histological techniques (29% of the module's qualification):

- Attitude and active participation in practical sessions (10%)
- Individual delivery of a report and questionnaire (45%)
- Image composition using the Photoshop program (45%).

2. Cell culture, fluorescence in situ hybridization and immunocytofluorescence (50% of the module's qualification):

- Attitude and active participation in practical sessions (10%)
- Delivery of a report with the results obtained through the application of these techniques (90%)

3.Confocal Laser Scanning Microscopy (14% of the module's qualification):

- Attitude and active participation in practical sessions (10%)
- Written exam (90%).

4. Chromosome identification: karyotype (7% of the module's note):

- Attitude and active participation in practical sessions (10%)
- Resolution of normal and altered karyotypes with the"Human Karyolab" program (40%)
- Resolution and delivery of karyotypes with anomalies, indicating the formula, clinical characteristics of the anomaly and the risk of having affected offspring (50%)

Recovery/ Retake process

The "Integrated Cytogenetics Laboratory" module, being eminently practical, does not allow the existence of recovery tests, unless, the responsible teacher of the block, with the approval of the module coordinator, can schedule a recovery.

To be eligible for the retake process, the student should have been previously evaluated in a set of activities equalling at least two thirds of the final score of the course. Thus, the student will be graded as "No Avaluable" if the weighting of all conducted evaluation activities is less than 67% of the final score.

"No Avaluable" qualification

Students will get the "No Avaluable" qualification when:

- Non-attendance at programmed sessions is greater than 20%.
- The weighting of all conducted evaluation activities is less than 67% of the final score.

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Cell culture, fluorescence in situ hybridization and immunocytofluorescence	50%	0	0	4, 3, 2, 5, 13, 8, 9, 17, 11, 14, 6, 7, 10, 15, 16
Chromosome identification: karyotype	7%	0	0	3, 11, 14, 6, 7, 16
Confocal Laser Scanning Microscopy	14%	0	0	4, 3, 13, 11, 14, 6, 7, 12, 16
Update in histological techniques	29%	0	0	4, 1, 13, 14, 7, 16

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

Adobe Photoshop

Microsoft office