Clinical Cytogenetics

Code: 42943
ECTS Credits: 6

2024/2025

Degree	Type	Year
4313782 Cytogenetics and Reproductive Biology	OT	0

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Teachers

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Teaching groups languages

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

Prerequisites

The study indicated for this Masters.

Objectives and Contextualisation

- Updating knowledge of the latest advances in cytogenetics and clinical genetics which offer an accurate diagnosis of hereditary diseases.
- Relating chromosomal alteration associated with the phenotype.
- Knowing the critical chromosomal regions associated with the most common hereditary diseases.
- Understanding the basis of genetic counseling and how to calculate risk offspring of inherited diseases affecting carriers of chromosomal alterations.
- Identify the advantages and disadvantages of each method of prenatal diagnosis (invasive and noninvasive).

Competences

- 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.
- 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.
- Interpret, resolve and report on clinical cases or scientific findings in the area of the master's degree.
- Respect ethical principles in one's work.
- 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. Develop technologies to be applied in genetics and clinical cytogenetics or in research in the public or private sector.
5. Evaluate the risk of affected offspring in carrier individuals and contribute to genetic counselling.
6. Identify and take into account the genetic changes involved in chromosome pathologies.
7. Integrate knowledge and use it to make judgements in complex situations, with incomplete information, while keeping in mind social and ethical responsibilities.
8. Interpret and diagnose human karyotype disorders.
9. Respect ethical principles in one's work.
10. Solve problems in new or little-known situations within broader (or multidisciplinary) contexts related to the field of study.
11. Use acquired knowledge as a basis for originality in the application of ideas, often in a research context.
12. Use and manage bibliography or ICT resources in the master's programme, in one's first language and in English.
13. Write articles or report scientific findings in the area of clinical cytogenetics.

Content

Unit 1: Clinical cytogenetics. Clinical consequences of germ and somatic abnormalities. Individuals mosaics. Frequency in population. Detection of chromosomal abnormalities in the population. Origin of numerical abnormalities and structural anafásica loss and non-disjunction. Chromosomal breakage.

Theme 2: Frequent chromosomal pathologies . General characteristics and associated clinical features. Deletion and trisomy viable human species. Autosomal chromosome abnormalities. Changes in sex chromosomes. Molecular. Regions criticism genotype-phenotype correlation.

Item 3: Structural alterations. Balanced alterations. Risk progeny in carriers. Translocations and invertions. Alterations frequently unbalanced. Marker chromosomes. Syndromes associated with microdeletions and microduplicacions.

Item 4: Genetic Counselling and Prenatal Genetic Diagnosis. Genetic counseling. Directions to perform prenatal diagnosis. Methods of Prenatal Diagnosis: invasive and noninvasive. Miscarriages in the first trimester.

Item 5: Introduction to the application of new technologies in the postnatal genetic study. The arrays-CGH and Optical Genomic Mapping, the new benchmarks of diagnostic technology and genetic analysis in the field of intellectual disability and congenital malformations.

Item 6: Speciality in Medical Genetics. Organization of Clinical Genetics at the state level. Access to the speciality. Medical Genetics Services . Database of Human Genetics online. Interrelation of inter-hospital services. Reference centers

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Identify alterations in the human karyotype and clinical consequences	8	0.32	1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13
Working together to resolve issues in genetic counseling and risk calculation in offspring affected by hereditary diseases	8	0.32	1, 2, 4, 5, 6, 7, 8, 10, 12
lectures	30	1.2	1, 2, 3, 6, 7, 8, 10, 11
Type: Supervised			
Written and Oral Work	4	0.16	1, 2, 3, 4, 7, 9, 10, 11, 12, 13
Type: Autonomous			
Deevelopment and integration of knowledge work	60	2.4	1, 2, 7, 9, 10, 11, 12, 13
autoevaluation	10	0.4	3, 4, 6, 7, 8, 10

1. Classes with theoretical support of ICT.
2. Self-evaluation exercises of the module contents.
3. Personal Work: each student has to work on a specific subject module (oral presentation and manuscript)

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

Continous Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Active participation in class discussions	10%	27	1.08	2, 3, 6, 7, 9, 10, 11
Presentation and defense cytogenetics work	60%	2	0.08	1, 4, 5, 11, 12, 13

Review test	30%	1	0.04	1, 5, 6, 8

Evaluation System

The competences of this course will be evaluated through participation in class, preparation of works and i *mplementation review*.

The evaluation is individual:

1. Continuous Assessment for active participation and discussions in class: 10% final.

2. Examination test (options 4/1 correct, penalty 1/3): 30% of final.

3. Presentation and defense of a work related on cytogenetics: 60% final.

- Oral presentation. Its important to answer the questions raised by colleagues and teachers: 30% of final grade.

- Written presentation. It will be necessary for the work to show the improvement comments made during the oral presentation : 30% final.

4. Recovery exam (50% text- 50% exposition and written presentation).

Remember that class attendance is mandatory.

Bibliography

Books:

- Genetics and Genomics in Medecine. Strachan et al. (2015). Ed Garland Science.

- Human Genetics and Genomics. Korf BR (2012). Ed Willey-Blackwell, 4th ed.

- Genética Humana. Fundamentos y aplicaciones en Medicina. Solari AJ (2011). Ed Médica Panamericana, 4ª ed.

- Genética Médica. Jorde LB (2016). Ed Elsevier, 5ª ed.

- New Clinical Genetics. Read and Donnai (2010). Ed Scion Publishing Ltd, 2nd ed.

- Human Genetics: Concepts and applications. Lewis R (2010). Ed McGraw-Hill International, 9nd ed.

- Elementos de Genética Médica. Emery et al. (2009). Ed. Elsevier,13ª ed.

- Genética en Medicina. Thompson and Thompson (2016). Ed Elsevier, 8ª ed.

- Human chromosomes. Miller and Therman (2001). Ed Springer, 4th ed.

- Genetics of complex desease. Donalson et al (2016). Ed Garland Science.

- Chromosome abnormalities and Genetic Counseling. Gardner and Sutherland (2011) Ed Oxford University Press.

- Human cytogenetics : constitutional analysis : a practical approach. Rooney and Czepulkowski (2001) Ed Oxford University Press (3rd ed).

- ISCN. An International System for Human Cytogenetic Npmenclature (2020) McGowan-Jordan, Simons and Schmid (2016). Ed Karger.

- Cancer Cytogenetics: Chromosomal and Molecular Genetic Abberations of Tumor Cells. Felix Mitelman (2011). Ed Wiley Blackwell.

PubMed http://www.kumc.edu/gec/prof/cytogene.html

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

Genetics Home Reference http://ghr.nlm.nih.gov/ghr/page/Home

Cytogenetic Resources http://www.kumc.edu/gec/prof/cytogene.html

University of Wisconsin http://www.slh.wisc.edu/wps/wcm/connect/extranet/cytogenetics

Additional documentation is available on the Virtual Campus

Software

Application software is the set of programs that allow document management, data processing, digital image retouching, Internet browsing ... etc. The application software can be grouped into the following sections:

Word processors: allow the creation of documents with the integration of text, data and images. Examples: Word, WordPad ...
Databases: they allow the processing of large amounts of information and facilitate subsequent consultation. Example: Access.
Spreadsheets: allow the processing of data, but in this case numerical, and the performance of mathematical calculations. Example: Excel.
Communication programs: allow communication through computer networks. You can browse the Internet, send an email ... Examples: IE, Browser, Messenger, Outlook, Eudora ...
Graphic design: they allow the realization and digital treatment of drawings, plans and photographs. Examples: AutoSketch (technical drawing), Paint (drawing), Paint Shop Pro (image processing) ...
Multimedia creation programs: allow you to integrate text, graphics, sound and animations, and create interactive documents. Examples: Flash, PowerPoint ...
... and the list of programs could go on with file compressors, MP3 players ... and a long and so on.

Diagnostic software:
*I*s the set of programs that allow you to configure and check the correct operation of all the hardware elements of a computer system.

Language list

Name	Group	Language	Semester	Turn
(PAULm) Classroom practices (master)	1	Catalan	first semester	morning-mixed
(TEm) Theory (master)	1	Catalan	first semester	morning-mixed