Universitat Autònoma de Barcelona

Clinical Cytogenetics 2016/2017

Code: 42943
ECTS Credits: 6

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

Contact

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Use of languages

Principal working language: catalan (cat)

Teachers

Vicenç Català Cahís

Alberto Plaja Rustein

External teachers

Miriam Guitart Feliubadaló

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).

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.
- 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: 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

Methodology

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)

Activities

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

Evaluation

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 on cytogenetics: 60% final.

- Oral presentation. We will respond to the issues raised by peers and teachers: 30% of final grade.

- Written presentation. Will you collect the comments made during the oral presentation: 30% final.

Remember that class attendance is mandatory.

Evaluation activities

Title	Weighting	Hours	ECTS	Learning outcomes

Active participation in class discussions	10%	27	1.08	9, 6, 10, 2, 7, 3, 11
Presentation and defense cytogenetics work	60%	2	0.08	1, 5, 4, 13, 11, 12
Review test	30%	1	0.04	1, 5, 6, 8

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 (2016) McGowan-Jordan, Simons and Schmid (2016). Ed Karger.

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