Clinical Cytogenetics

Code: 42943
ECTS Credits: 6

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<th>Degree</th>
<th>Type</th>
<th>Year</th>
<th>Semester</th>
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<td>4313782 Cytogenetics and Reproductive Biology</td>
<td>OT</td>
<td>0</td>
<td>1</td>
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Contact

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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.


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

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

Evaluation System

The competences of this course will be evaluated through participation in class, preparation of works and implementation 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. 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

<table>
<thead>
<tr>
<th>Title</th>
<th>Weighting</th>
<th>Hours</th>
<th>ECTS</th>
<th>Learning outcomes</th>
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<tbody>
<tr>
<td>Active participation in class discussions</td>
<td>10%</td>
<td>27</td>
<td>1.08</td>
<td>9, 6, 10, 2, 7, 3, 11</td>
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<tr>
<td>Presentation and defense cytogenetics work</td>
<td>60%</td>
<td>2</td>
<td>0.08</td>
<td>1, 5, 4, 13, 11, 12</td>
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Bibliography

Books:


Orphanet [http://www.orpha.net/consor/cgi-bin/home.php?Lng=ES](http://www.orpha.net/consor/cgi-bin/home.php?Lng=ES)


Additional documentation is available on the Virtual Campus