

26786 - Medical genetics

Información del Plan Docente

Academic Year	2017/18
Faculty / School	104 - Facultad de Medicina
Degree	304 - Degree in Medicine
ECTS	5.0
Year	5
Semester	First semester
Subject Type	Optional
Module	---

1.General information

1.1.Introduction

1.2.Recommendations to take this course

1.3.Context and importance of this course in the degree

1.4.Activities and key dates

2.Learning goals

2.1.Learning goals

2.2.Importance of learning goals

3.Aims of the course and competences

3.1.Aims of the course

3.2.Competences

4.Assessment (1st and 2nd call)

4.1.Assessment tasks (description of tasks, marking system and assessment criteria)

5.Methodology, learning tasks, syllabus and resources

5.1.Methodological overview

5.2.Learning tasks

5.3.Syllabus

Program

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Human genome:

1. Genes: Organization, expression control, segregation.
2. Individual Variability: mutations and polymorphisms.

Diseases of genetic etiology

1. Classification.
2. Chromosomopathies
3. Monogenic diseases. Factors that interfere with Mendelian patterns. Dynamic mutations. Imprinting.
4. Mitochondrial diseases
5. Diseases of complex inheritance
6. Determination / sexual differentiation
7. Developmental Genetics
8. Pathologies by genetic instability. Cancer Genetics
9. Genetic counseling. Carrier detection
10. Gene Therapy

Practical Classes

They will be held at the Laboratory Practices and students will acquire the information about:

Nomenclature of Medical Genetics: Genes, mutations, polymorphisms, chromosomal alterations

The methodological basis of cytogenetic analysis and molecular genetic analysis

Protocols and diagnostic techniques in medical genetics and interpretation of: Cytogenetic and molecular diagnose

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Clinical Seminars

They will be conducted in small groups, exposing and discussing: Clinical cases, genetic counseling, Databases in Medical Genetics

5.4.Course planning and calendar

5.5.Bibliography and recommended resources

- 0 - Genética en Medicina Thompson & Thompson, Nussbaum RL, McInnes RR, Willard HF, Hamosh A. Elsevier Masson, 7ª edición, 2008
- Genética Humana. Fundamentos y aplicaciones en Medicina. Solari, AJ. Ed Panamericana, 4ª edición, 2011
- Genética. Texto y Atlas Passarge E. Editorial Médica Panamericana, 3ª edición, 2009
- Genética humana. Stracham T, Read AP. McGrawHill, 3ª edición, 2006
- Nueva Genética clínica. Read A, Donnai D. Ed Omega, 2008
- Biologie Moléculaire et Médecine J.C. Kaplan & M.Delpech. Ed. Flammarion (Médecine-Sciences), 3ª edición, 2007
- Elementos de Genética médica. Turnpenny P, Ellard S. Ed Elsevier, 13ª edición, 2009

PubMed

<http://www.ncbi.nlm.nih.gov/pubmed>

Online Mendelian Inheritance in Man (OMIM)



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<http://www.ncbi.nlm.nih.gov/omim>

Geneclinics, GeneTests

www.geneclinics.org

<https://www.genetests.org/>

Human Genome Project Information

<http://ghr.nlm.nih.gov/handbook/hgp>

Biblioteca Universitaria Zaragoza

<http://biblioteca.unizar.es/>