

26786 - Medical genetics

Información del Plan Docente

Academic Year	2016/17
Academic center	104 - Facultad de Medicina
Degree	304 - Degree in Medicine
ECTS	5.0
Course	5
Period	First semester
Subject Type	Optional
Module	---

1.Basic info

1.1.Recommendations to take this course

1.2.Activities and key dates for the course

2.Initiation

2.1.Learning outcomes that define the subject

2.2.Introduction

3.Context and competences

3.1.Goals

3.2.Context and meaning of the subject in the degree

3.3.Competences

3.4.Importance of learning outcomes

4.Evaluation

5.Activities and resources

5.1.General methodological presentation

5.2.Learning activities

5.3.Program

Program

Human genome:

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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

Clinical Seminars

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

5.4.Planning and scheduling

5.5.Bibliography and recommended resources

- o - Genética en Medicina Thompson & Thompson, Nussbaum RL, McinnesRR, 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)

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

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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/>