

69004 - Basic concepts in Genetics. Diagnosis techniques. Gene appraisal and therapy. Treatment of metabolic illnesses

Información del Plan Docente

Academic Year	2017/18
Faculty / School	104 - Facultad de Medicina
Degree	461 - Master's in Genetic, nutritional and environmental growth and development conditions
ECTS	1.0
Year	1
Semester	Annual
Subject Type	Compulsory
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

This course has no face-to-face sessions, it is done via the virtual platform Moodle. The course contents include original works and/or explanatory presentations related to topics of great social and medical significance.

Although the students have access to the materials on the platform, teachers will be available for queries and doubts via email, messages and chats.

69004 - Basic concepts in Genetics. Diagnosis techniques. Gene appraisal and therapy. Treatment of metabolic illnesses

5.2.Learning tasks

5.3.Syllabus

The course will address the following topics:

- Topic 1. General Concepts of Clinical Genetics. Types of inheritance
- Topic 2. Clinical Genetics History. Importance of the family tree
- Topic 3. Introduction to Dysmorphology
- Topic 4. Genetic Counseling
- Topic 5. Introduction to Clinical Cytogenetics. Clinic and diagnosis of major chromosomal abnormalities
- Topic 6. Basics of Molecular Genetics

5.4.Course planning and calendar

This course takes place during the months of May and June of the academic year.

Submission dates of assignments are informed at the beginning of the course through a message/announcement of the teacher.

5.5.Bibliography and recommended resources

Thompson & Thompson "Genetics in Medicine". Nussbaum RL, McInnes RR,Willar HF Eds. 8th ed. Elsevier, Philadelphia, 2015.

- Emery & Rimoin "Principles and Practice of Medical Genetics". Rimoin DL, Connor JM, Pyeritz RE, Korf BR Eds. 6th ed. Elsevier, San Diego, 2013.

- Smith's Recognizable Paterns of Human Malformation. Jones KL, Jones MC, Del Campo M Ed. 7th ed. Saunders, Philadelphia, 2013.

- The Metabolic and Molecular Bases of Inherited Disease. Scriver CR, Sly WS, Childs B, Beaudet AL, Valle D, Kinzler KW, Vogelstein B Eds. 8th ed. McGraw-Hill, New York, 2001.

Enlaces recomendados

www.orpha.net

www.omim.org

www.genetests.org

www.genereviews.org