26786 - Medical genetics

Información del Plan Docente

Academic Year	2018/19
Subject	26786 - Medical genetics
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.Aims of the course

1.2.Context and importance of this course in the degree

1.3.Recommendations to take this course

2.Learning goals

2.1.Competences

2.2.Learning goals

2.3.Importance of learning goals

3.Assessment (1st and 2nd call)

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

4.Methodology, learning tasks, syllabus and resources

4.1.Methodological overview

Methodological overview

The methodology followed in this course is oriented towards achievement of the learning objectives:

-Obtain and elaborate a clinical history with relevant information.

-Perform a physical examination and a mental health assessment.

-Have the capacity to make an initial diagnosis and establish a reasonable strategy of diagnosis.

-Establish the diagnosis, prognosis and treatment, applying principles based on the bestinformation available and on conditions of clinical safety.

-Know how to use the sources of clinical and biomedical information available, and value them critically in order to obtain, organise, interpret and communicate scientific and sanitary information.

In the theoretical lessons the teacher will expose, through master class, the most important concepts and contents in a structured way, to obtain of the knowledge and skills that the students must acquire. Students are expected to participate actively in the class throughout the semester.

Classroom materials will be available via Moodle. These include a repository of the lecture notes used in class, the course syllabus, as well as other learning resources. Also, classroom practices, case studies, the group work and oral presentation will be encouraged.

4.2.Learning tasks

Learning tasks

The course includes 5 ECTS organized according to:

1.- Theoretical lessons: (2,5 ECTS) : 25 hours

2.- Práctical lessons: (2,5 ECTS) : 25 hours

Presentation of clinical cases :15 hours

Laboratory practices: 10 hours

4.3.Syllabus

Program

26786 - Medical genetics

Human genome:

1. Genes: Organization, expression control, segregation.

2. Individual Variability: mutations and polymorphisms.

Diseases of genetic etiology

1. Classification.

2. Chromosomopaties

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

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

Laboratory Practices

Molecular diagnosis techniques

Mutations analysis.

Analysis of structural genetic variations.

Databases management

4.4.Course planning and calendar
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Planning of learning activities and calendar of key dates

Theoretical and practical classes: From the first week to the fourteenth week of the course

Programming classes: Two theoretical hours per week and two practical hours per week.

Initially these hours will be: Monday and Wednesday from 10am to 12pm.

The programming of the Laboratory Practices and corresponding Groups of Practices will be done once enrolled students

4.5.Bibliography and recommended resources

http://psfunizar7.unizar.es/br13/eBuscar.php?tipo=a