Universidad Zaragoza

Academic Year/course: 2023/24

26786 - Medical genetics

Syllabus Information

Academic year: 2023/24
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

Genetics in the field of medicine has evolved from phenotypic observation to personalized genetics with new diagnostic techniques, genomic editing and gene therapy, among others. For this reason, it is necessary in the development of the future physician's learning . Erasmus students can study at a distance.

Objectives

1.- To know the aetiology, pathogenesis and diagnostic procedures of the main genetic diseases.

2.- To be able to establish a diagnostic/prognostic action plan according to the needs of the patient and their family environment

3.- To know and know how to use the tools of genetic diagnosis.

4.- To know how to adequately attend the patient and/or applicant for genetic counselling and advice.

Its approach and objectives are aligned with Sustainable Development Goal 3 (SDG) of the United Nations 2030 Agenda, Health and Wellness

2. Learning results

The student, in order to pass this subject, must demonstrate the following results...

- To know the aetiology, pathogenesis and diagnostic procedures from the point of view of Genetics.
- To acquire the knowledge and skills to establish an action plan according to the needs of the patient and their family environment.
- To detect the hereditary component of a disease, distinguish between sporadic, familial and hereditary.
- To perform confirmatory diagnosis of genetic diseases
- To calculate the risk of disease transmission, identify individuals at risk and make presymptomatic diagnoses.
- To know the information provided by genetic diagnostic tests and techniques. To know how to interpret the corresponding reports.
- To determine the appropriate diagnostic tests in each case and interpret the results of these tests To manage the Human Genome Databases (GDB) and genetic computation tools.

The learning results are:

- Relevant in the daily professional practice of future physicians to:

1. Identify and diagnose diseases of genetic aetiology
2. Attend to the therapeutic demands of the patient
3. Provide access to information and genetic counselling for the patient

- Essential as a scientific basis for research oriented to the knowledge of the aetiology of human diseases and, consequently, to the design of effective therapies based on their physiopathology.

All this will contribute to:

- Adequately attend to the patient and/or applicant for genetic consultation To contribute to the prevention of genetic diseases

- Collaborate in the health education of the population regarding the early diagnosis of diseases of genetic aetiology and the importance of participating in early detection and carrier study programs.

3. Syllabus

BASIC CONCEPTS OF HUMAN GENETICS

- Human genome, genetic variation and its impact on health

- Types of genetic variations according to clinical impact and relevance of their analysis for clinical practice
- Monogenic and multifactorial pathologies
- Cancer genetics
- Mitochondrial genetics

CLINICAL GENETIC ANALYSIS STRATEGY

- Genetic analysis techniques currently used for disease diagnosis, NGS
- Genetic analysis strategies used in clinical practice.
- Gene panels and case studies
- Exome analysis and case studies
- Genome analysis and case studies

GENETIC COUNSELING AND ADVICE

OUTLOOK AND FUTURE OF GENOMICS IN CLINICAL PRACTICE

4. Academic activities

Learning activities of the subject will be carried out:

The theoretical knowledge necessary for the understanding and learning of the genetic aetiology of human disease will be presented in the face-to-face classes.

Students will discuss and solve clinical cases previously selected by the teacher that deal with the main human pathologies with a genetic component.

Students will perform practical laboratory sessions that will familiarize them with the interpretation and clinical application of the main diagnostic techniques, both cytogenetic and molecular genetics.

The subject is structured in:

Theoretical sessions (26 hours/student) and **practical sessions** (20 hours/student)

Students will be informed about the risks and must sign a commitment to comply with the work and safety standards in order to be able to perform them. http://uprl.unizar.es/estudiantes.html".

5. Assessment system

THEORETICAL CONTENTS.

In order to pass the theoretical contents a minimum grade of 5 points out of 10 is required. This grade will be obtained through the activities of the subject in the Continuous Evaluation or a global exam of all the theoretical contents of the subject if the student does not opt for continuous evaluation.

PRACTICAL CONTENTS

The positive evaluation of the practices contents is mandatory to pass the subject. A minimum grade of 5 out of 10 will be required to pass and will be **obtained through attendance and effective participation in the respective practices**

A. Laboratory Practices.

B. Practical Cases.

In these activities, the teacher will tutor students

The final exam, for those who do not opt for continuous evaluation, will consist of short questions of development and resolution of a practical case, assessing the ability to express, synthesize and relate concepts.

Percentages of the final grade obtained by the student Theory Contents =60%

Practices contents = 40%

Erasmus students taking the subject by distance learning may opt for continuous assessment or a final exam