

26786 - Medical genetics

Syllabus Information

Academic Year: 2022/23

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

The course and its expected results respond to the following approaches and objectives:

Objectives

- 1.- Know the etiology, pathogenesis and diagnostic procedures of the main genetic diseases.
- 2.- Be able to detect the genetic component of a disease, with special attention to the genetic risk of diseases of complex inheritance.
- 3.- Be able to calculate the risk of transmission of a disease, identify individuals at risk and perform presymptomatic diagnoses.
- 4.- Be able to establish a diagnostic / prognostic action plan according to the needs of the patient and her family environment. Give advice and genetic counselling.
- 5.- Know how to interpret and assess the information provided by genetic diagnostic techniques in the context of human clinical practice.
- 6.- Know how to use clinical genetic databases and those of the Human Genome (GDB) and genetic calculation tools.
7. Know the new diagnostic tools for personalized Genetics

These approaches and objectives are aligned with the following Sustainable Development Goals (SDGs) of the United Nations 2030 Agenda (<https://www.un.org/sustainabledevelopment/es/>), in such a way that the acquisition of the results of Learning the subject of Medical Genetics provides training and competence to contribute to some extent to its achievement.

These approaches and objectives are in line with the following Sustainable Development Goals (SDGs) of the United Nations 2030 Agenda (<https://www.un.org/sustainabledevelopment/es/>), so that the acquisition of the learning outcomes of the subject provides training and competence to contribute to some extent to their achievement.

The objectives to be developed will be:

- ? Goal 3: Health and well-being.
- ? Goal 4: Quality education.
- ? Goal 5: Gender equality.

1.2. Context and importance of this course in the degree

Genetics in the field of Medicine has evolved from phenotypic observation to genotypic diagnosis and personalized medicine; from syndromic characterization to molecular pathology and genetic etiology.

Current diagnostic tests allow the detection of both affected and asymptomatic carriers, of hitherto unknown variants and those of uncertain significance, which may or may not be pathogenic.

For this reason, Medical Genetics is a current need in the development of knowledge, diagnosis, treatment and prevention of human diseases and the learning of this subject is a priority in the good practice of medical professionals trained to:

- 1) Recognize and diagnose pathologies of genetic etiology.
- 2) Know and identify the genetic predisposition factors involved in human pathology.
- 3) Assess changes in genetic parameters at the molecular and cellular level and interpret their clinical impact.
- 4) Adequate care for the patient and/or applicant for genetic counseling.

1.3. Recommendations to take this course

To facilitate the learning and study of the contents corresponding to Medical Genetics, students must have acquired the knowledge of the preceding subjects of Module I (Morphology, Structure and Function of the Human Body), Module II (Diagnostic and Therapeutic Procedures) and Module III (Human Clinical Training)

2. Learning goals

2.1. Competences

BASIC:

CB1 - Students have demonstrated possession and understanding of knowledge in an area of ??study that is based on general secondary education, and is usually found at a level that, although supported by advanced textbooks, also includes some aspects involving knowledge from the forefront of their field of study

CB2 - That students know how to apply their knowledge to their work or vocation in a professional way and possess the skills that are usually demonstrated through the elaboration and defense of arguments and the resolution of problems within their area of ??study

CB3 - That students have the ability to gather and interpret relevant data (normally within their area of ??study) to make judgments that include a reflection on relevant issues of a social, scientific or ethical nature

CB4 - That students can transmit information, ideas, problems and solutions to both a specialized and non-specialized audience

CB5 - That students have developed those learning skills necessary to undertake further studies with a high degree of autonomy

SPECIFICS:

Adequately attend to the patient and/or claimant of genetic consultation

Contribute to the prevention of genetic diseases

Collaborate in the health education of the population regarding the early diagnosis of diseases of genetic etiology and the importance of participating in early detection programs and carrier studies

TRANSVERSAL

As a reference for the selection, the skills collected in the Tunning project can be used:

a. INSTRUMENTAL

1. Capacity for analysis and synthesis
2. Information management capacity
3. Troubleshooting
4. Decision making

b. PERSONAL

9. Teamwork
12. Skills in interpersonal relationships
13. Recognition of diversity and multiculturalism
14. Critical reasoning
15. Ethical commitment

c. SYSTEMIC

16. Autonomous learning
17. Adaptation to new situations
18. Motivation for quality

2.2. Learning goals

Know the etiology, pathogenesis and diagnostic procedures from the point of view of Genetics.

Acquire the knowledge and skills to establish an action plan according to the needs of the patient and her family environment.

Detect the hereditary component of a disease, distinguish between sporadic, family and hereditary.

Carry out the confirmation diagnosis of genetic diseases

Calculate the risk of transmission of a disease, identify individuals at risk and perform presymptomatic diagnoses.

Know the information provided by genetic diagnostic tests and techniques. Know how to interpret the corresponding reports.

Determine the appropriate diagnostic tests in each case and interpret their results

Manage the Human Genome (GDB) databases and genetic calculation tools.

2.3. Importance of learning goals

The learning goals are:

Relevant to the daily professional practice of future physicians to:

- 1) Identify and diagnose diseases of genetic etiology.
- 2) Attend the therapeutic demands of the patient.
- 3) Provide to the patient access to the information and advice about genetic counselling

Essential as scientific basis of those investigations oriented to the knowledge of the etiology of human diseases and consequently to the design of effective therapies based on their pathophysiology.

All this will contribute to:

Adequate attention to the patient and/or applicant for genetic consultation.

Contribute to the prevention of genetic diseases.

Collaborate in the health education of the population regarding the early diagnosis of diseases of genetic etiology and the importance of participating in early detection programs and the study of carriers.

3. Assessment (1st and 2nd call)

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

The student must demonstrate that he/she has achieved the expected learning results through the following evaluation activities

THEORETICAL CONTENTS.

In order to pass the theoretical contents a minimum score of 5 points out of 10 is required, which each student will obtain by means of a final written exam or global exam of all the theoretical contents of the subject.

This exam will consist of a written test with short multiple-choice questions in which the correct answer will be justified by means of a brief text. The capacity of comprehension and correlation of concepts, clarity of expression and schematization will be valued.

It will take place during the examination period of the official exams.

PRACTICAL CONTENTS

The positive evaluation of the Practical contents is mandatory to pass the course. In order to pass it, a minimum grade of 5 points out of 10 will be required, which will be obtained by means of the attendance and effective participation in the respective Practicals.

A) Laboratory Practices. Students with two or more absences will have to take an exam.

B) Clinical Seminars. Students must prepare a clinical case or seminar to be presented to the rest of the class:

This seminar will be individual or prepared in small groups of students.

Its content will be focused on topics related to the objectives and contents of the course.

The participation in the seminars includes searching for the appropriate material, preparing, presenting the seminar to the whole class, defending what has been presented, and so on.

The seminars will be posted in the ADD (text, images).

This activity will be graded individually, evaluating the preparation of the seminar, the clarity in the transmission of concepts and the expository capacity.

In this activity, students will be tutored by the professor.

Percentages over the final grade obtained by the student

Theory contents = 60%.

Practical Contents = 40%, respectively broken down into: 1/3 for Laboratory Practicals and 2/3 for Clinical Seminars.

4. Methodology, learning tasks, syllabus and resources

4.1. Methodological overview

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

- Obtain and elaborate on 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 best information 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 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

The course includes 5 ECTS organized according to:

- 1.- Lectures: (2,5 ECTS): 25 hours
- 2.- Practice sessions: (2,5 ECTS): 25 hours
 - Presentation of clinical cases: 15 hours
 - Laboratory practices: 10 hours

4.3. Syllabus

The course will address the following topics:

Human genome

1. Genes: organization, control of expression, segregation.
2. Individual variability: mutations and polymorphisms. Significance in Medicine

Diseases of genetic etiology

1. Classification.
2. Cytogenetics.
3. Monogenic diseases. Factors interfering with Mendelian patterns. Dynamic mutations. Imprinting.
4. Epigenome and disease
5. Mitochondrial diseases
6. Multifactorial or complex inheritance diseases.
7. Sexual differentiation. Reproductive genetics and neonatal screening.
8. Pathologies by genetic instability. Cancer genetics

Genetics in medicine

9. Genetic diagnosis
10. Genetic counseling. Carrier detection
11. Gene therapy

Technology and Genetics

12. Genetic analysis techniques. Sequencing, qPCR, RT-PCR, NGS, MedIP-seq. BigData

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

Course planning and calendar

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 10 am. to 12 pm.

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

GLOBAL ASSESSMENTS <https://medicina.unizar.es/quinto-curso#horario9>

4.5. Bibliography and recommended resources

<http://psfunizar10.unizar.es/br13/egAsignaturas.php?codigo=26786>