Universidad Zaragoza

**Academic Year/course: 2024/25**

# 32306 - Medical Genetics

## Syllabus Information

**Academic year:** 2024/25
**Subject:** 32306 - Medical Genetics
**Faculty / School:** 104 - Facultad de Medicina
229 - Facultad de Ciencias de la Salud y del Deporte
**Degree:** 649 - Degree in Medicine
650 - Degree in Medicine
**ECTS:** 3.0
**Year:** 1
**Semester:** Second semester
**Subject type:** Compulsory
**Module:**

## 1. General information

Human Genetics studies the inheritance and variation of biological traits in humans, constituting one of the fundamental pillars of Biomedicine today.

The course provides a global and integrative view of applying genetics to the study of inheritance in the human species.

It is taught in the second semester of the first year of the Medicine degree. Students will acquire advanced knowledge about the human genome, epigenetics, gene expression regulation, inheritance patterns, cytogenetics, genetic etiology of diseases, and cancer genetics. These foundational concepts will serve as a basis for other subjects in the degree program. Additionally, students will gain a comprehensive understanding of the morphology, structure, and function of the human body across different life stages.

## 2. Learning results

Upon completing this course, students will be able to:

1. Apply basic techniques commonly used in the genetics laboratory.
2. Understand scientific texts and create review papers on Human Genetics and genetic diseases.
3. Compare techniques and methods used in genetic diagnosis.
4. Demonstrate basic research skills.
5. Exhibit a critical, creative, and research-oriented perspective in professional activities.
6. Describe the organization, evolution, interindividual variation, and expression of the human genome.
7. Explain chromosomal abnormalities in humans and evaluate their consequences.
8. Describe the molecular basis of DNA mutation and repair.
9. Highlight the importance of research in the field of genetics.
10. Explain mechanisms of genetic material transmission.
11. Formulate hypotheses and critically evaluate information for problem-solving using the scientific method.
12. Identify the genetic bases of major diseases with a genetic component.
13. Understand genetic concepts and terminology, and consult scientific literature related to human genetics.
14. Identify epigenetic factors involved in gene expression control.
15. Interpret results from scientific projects.
16. Analyze scientific publications, solve problems, and work with case examples in the field of genetics.
17. Relate genetic dysfunction to pathological phenotypes.

## 3. Syllabus

### Theory

1. **Human Genome**: Structure and levels of organization. Genes. Repetitive DNA.
2. **Molecular Genetics**: Mutations, polymorphisms, and inheritance models. Diagnostic techniques.
3. **Cytogenetics**: Chromosomes, mitosis, meiosis. Diagnostic techniques. Chromosomal disorders.
4. **Epigenetics**: General gene regulation. Telomerase. Epigenetic elements. Diagnostic techniques.
5. **Cancer**: Study of molecular and genetic mechanisms in cancer.
6. **Genetic Counseling**: Diagnostic techniques (prenatal diagnosis).

### Practical Sessions

1. **Introduction to Genetic Laboratory**: DNA extraction.
2. **Paternity Testing**
3. **Hereditary Traits and Pedigrees**
4. **Blood Groups and Sex Chromatin**: Determination of ABO and Rh blood groups.
5. **Karyotypes**

### Practical Cases

1. **Problem-Solving Sessions** (3 sessions per student)
2. **Scientific Seminars** (4 sessions per student)

## 4. Academic activities

# Theoretical Classes (18h)

In theoretical classes, a lecture format will be used, lasting for 1 hour. The professor will make the necessary material accessible on the virtual teaching support platform, Aula Virtual, to ensure proper class follow-up.

# Practical Sessions (12h)

1. **Laboratory Practices**: These practical classes last for 1 hour and are mandatory.
2. **Problem-Solving Classes**: In these 1-hour sessions, concepts introduced in theory classes will be reinforced.
3. **Seminars, Conferences, and Other Activities**
4. **Personalized Tutoring**

## 5. Assessment system

The evaluation of the course includes:

1. **Final Theoretical-Practical Exam**
2. **Obligatory Activities for Continuous Assessment**

Students must demonstrate comprehension and mastery of the theoretical and practical knowledge covered in the course, including appropriate concepts and terminology.

To pass the course, students must independently pass each component contributing to the final grade. Each activity is graded on a scale of 0 to 10. A score below 5 in the written theoretical-practical exam results in failing the course. Similarly, a score below 5 in any section leads to failure.

## 6. Sustainable Development Goals

3 - Good Health & Well-Being
4 - Quality Education