1. OVERVIEW

Subject Area	GENETICS
Degree	MEDICINE
School/Faculty	BIOMEDICAL AND HEALTH SCIENCES
Ac. Year	1º
ECTS	4
Type	COMPULSORY
Language(s)	SPANISH
Delivery Mode	ON CAMPUS
Semester	2º

2. INTRODUCTION

Genetics is taught in the first year of the Degree in Medicine in the second term. This subject will provide students with the knowledge required for understanding the genetic make-up of many diseases. Students will study the organisation of the human genome in great detail, as well as the main reported mutations which have an effect on human health. We analyse the origin of genetic variation on both an individual and population level. We will pay particular attention to the use of genetics in medical practice through knowledge and the use of genetic databases, as well as everything associated to diagnosis and genetic counselling. Students will also learn about the opportunities the latest genetic developments are offering in the field of medicine such as pharmacogenetics, gene therapy, genome editing or the hereditary basis of oncologic diseases.

The general contents are as follows:
- Fundamentals of human genetics.
- Molecular bases of monogenic, polygenic and multifactorial diseases.
- Genetic disorders.
- Population genetics.
- Basic analysis techniques for genetic studies.

3. SKILLS AND LEARNING OUTCOMES

Key skills (CB, as per the Spanish acronym):

- CB1: Students have demonstrated possession and understanding of knowledge in a study area that builds on general secondary education, and is typically at a level that, while supported by advanced textbooks, also includes aspects that involve knowledge from the forefront of their field of study.
- CB2: Students can apply their knowledge to their work professionally and possess the necessary skills, usually demonstrated by forming and defending opinions, as well as resolving problems within their study area.

GENERAL SKILLS (CG, as per the Spanish acronym):

- CG.7: Understand and recognise the normal structure and function of the human body. This includes studies of molecules, cells, tissue, organs and systems in the different stages of life.
- CG.11: Understand and recognise the effects of growth, development and ageing on the individual and the social environment.

TRANSVERSAL SKILLS (CT, as per the Spanish acronym):

- CT1: Communication: ability to engage in active listening, ask questions and respond in a clear and concise way, as well as to effectively express ideas and concepts. This includes concise and clear written communication.
- CT6: Problem solving: ability to solve an unclear or complex issue or situation which has no established solution and requires skill to reach a conclusion.

SPECIFIC SKILLS (CE by its acronym in Spanish):

- CE1.1.2: Understanding the fundamentals of human nutrition. Cell communication. Excitable membranes. The Cell Cycle. Cellular differentiation and proliferation. Gene expression, information and regulation. Heredity. Embryo development and organogenesis.
- CE1.2.2: Using basic laboratory techniques and materials. Interpreting a normal analysis. Using macroscopic, microscopic and imaging techniques to recognise the morphology and structure of tissue, organs and systems. Performing functional tests and determining vital signs and how to interpret them. The basic physical examination.

LEARNING OUTCOMES (RA, as per the Spanish acronym):

- Understand the basic concepts of human genetics.
- Understand the molecular bases of monogenic diseases.
- Recognise the genetic make-up of polygenic and multifactorial diseases.
- Establish the relationship between the structure and function of genes and their involvement in the development of genetic disorders.
- Understand the importance of population genetics studies and their relationship with the study of genetic disorders.
- Be aware of the contribution of new developments in molecular genetics research and their use in clinical practice.
- Solve genetic issues when they are part of clinical cases.
- Understand and experience the basic analysis techniques for genetic studies.

The following table shows how the skills developed in the course match up with the intended learning outcomes:

Skills	Learning outcomes
CB1, CG.B.7, CG.B.11, CE2	Understand the basic concepts of human genetics.
CB1, CB2, CG.B.11, CE2	Understand the molecular bases of monogenic diseases.
CB1, CB2, CG.B.7, CT1, CT6, CE4	Recognise the genetic make-up of polygenic and multifactorial diseases.
CB2, CG.B.7, CG.B.11, CT1, CE2	Establish the relationship between the structure and function of genes and their involvement in the development of genetic disorders.

CB1, CB2, CG.B.11, CT6, CE2	Understand the importance of population genetics studies and their relationship with the study of genetic disorders.
CB2, CG.B.7, CT6, CE4,	Be aware of the contribution of new developments in molecular genetics research and their use in clinical practice.
CB2, CE4, CT1, CT6	Solve genetic issues when they are part of clinical cases.
CT6, CE2, CE4	Understand and experience the basic analysis techniques for genetic studies.

4. CONTENTS

UNIT 1. HUMAN GENOME

1.1. Nuclear genome

1.2. Mitochondrial genome

UNIT 2. GENE VARIATIONS IN THE HUMAN GENOME I: RECOMBINATION

2.1. Genetic recombination in meiosis

2.2. Somatic recombination in immunoglobulin genes

UNIT 3. GENE VARIATIONS IN THE HUMAN GENOME II: MUTATION

3.1. Mutations in the nuclear genome

3.2. Numerical and structural chromosomal alterations

3.3. Gene mutations and their consequences

3.4. Mutations in the mitochondrial genome

UNIT 4. MENDELIAN INHERITANCE

4.1. Mendelian bases of inheritance

4.2. Types of inheritance

UNIT 5. CANCER INHERITANCE

5.1. Oncogenes and tumour suppressor genes

5.2. Epigenetics and cancer

UNIT 6. PHARMACOGENETICS

6.1. Genes and enzymes of pharmaceutical metabolism phase I

6.2. Genes and enzymes of pharmaceutical metabolism phase II

UNIT 7. GENETIC LINKAGE

7.1. Linkage studies in humans

7.2. Localisation and identification methods for genes

UNIT 8. INHERITANCE LINKED TO SEX

8.1. Recessive and dominant inheritance pathologies linked to the X-chromosome

8.2. Gene dosage compensation

UNIT 9. QUANTITATIVE INHERITANCE

9.1. Heritability, variance, phenotype distribution

9.2. Multifactorial diseases

UNIT 10. GENE VARIATION IN HUMAN POPULATIONS

10.1. Populations in equilibrium: Hardy-Weinberg principle

10.2. Ethnic differences in the frequency of genetic disorders

UNIT 11. GENETICS IN MEDICAL PRACTICE

11.1. Retrospective and prospective diagnosis

11.2. Gene counselling

UNIT 12. TREATMENT OF HEREDITARY DISEASES: GENE THERAPY

12.1. Gene therapy against cancer
12.2 Genome editing systems

5. TEACHING/LEARNING METHODS

The types of teaching/learning methods are as follows:

- Problem-based learning: Presentation of problems, reorganising into small groups, literature analysis, analysis of scientific texts and documents, symposiums and presentations, directed debates, specialised individual and collective tutorials, and reaching a consensus.
- Case studies and problem solving: approach and solving cases and problems either as an individual or in small groups.
- Specialised seminars: literature research and debate on scientific data in small groups.
- Lectures: Classroom presentations by the lecturer on basic theory, encouraging debate and student participation.
- Learning based on specific laboratory teaching.

6. LEARNING ACTIVITIES

The types of learning activities, plus the amount of time spent on each activity, are as follows:

Learning activity	Number of hours
THEORY/PRACTICAL LEARNING ACTIVITIES	41
DIRECTED LEARNING ACTIVITIES	8
SELF-STUDY	40
TUTORIALS	8
KNOWLEDGE TESTS	3
TOTAL	**100**

7. ASSESSMENT

The assessment methods, plus their weighting in the final grade for the course, are as follows:

Assessment system	Weighting
Practical content (activities and laboratory practice)	30%
Objective tests	70%

The assessment of the theory part of the subject (70%) includes evaluation of theoretical content (50%) and skills and knowledge acquired during the practical subject activities (20%). The assessment of the practical part (30%) includes: problem solving, approaches and case solving, flipped classrooms, laboratory work, managing databases, etc. It also includes an assessment of the student's attitude.

On the Virtual Campus, when you open the subject area, you can see all the details of your assessment activities and the deadlines and assessment procedures for each activity.

BIBLIOGRAPHY

The recommended bibliography is indicated below:

- Gallego FJ, Fernández-Santander A. "Genómica y Proteómica". Ed. Sintesis, 2019.
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