Elective Subject

(Academic Course 2024-2025)

Subject title**: GENETICS APPLIED TO HEALTH SCIENCES**

Code: **806043**
Subject: **Elective**
Responsibility Center: **Faculty of Nursing, Physiotherapy and Podiatry**
Credits**: 3 ECTS**
Number of places offered: **40**

	Total		Theory	Seminars	Practices	Others
Classroom activities	20		13		7	
e-learning	4					4
Total	24		13		7	4

Course schedule: of the course (semester, day and schedule): First semester. Monday and Tuesday from 2:30 p.m. to 3:30 p.m. (possibility of adaptation)

STUDENT PROFILE (University degrees for which they are offered, if applicable)

Nursing, Physiotherapy and Podiatry

BRIEF DESCRIPTOR

Human Genetics plays an increasingly relevant role in health practice. Their study provides the student with basic concepts for a more precise understanding of the etiology of many pathological situations, of the most appropriate therapy or treatments and of the pertinent preventive actions.

Our genetic substrate plays a crucial role in our health and in our response to health care.
Health professionals must have basic genetic training for comprehensive health care.

The contents of this program include the progress made in recent years mainly focused on its clinical application.

OBJECTIVES

GENERALS
- Understand the importance of the role of genetics in health and disease.
- Provide students with the essential knowledge in human genetics that allows them to understand the physiopathology and the development of autonomous learning skills in this discipline.

SPECIFICS
- Know the characteristics of the human genome, chromosomes and mechanisms of genetic information maintenance between generations.
- Understand and interpret the main inheritance mechanisms of human characters.
- Understand the genetic basis of monogenic, chromosomal, multifactorial and mitochondrial disorders, as well as their clinical manifestations.
- Know the advances in the diagnosis and treatment of the diseases with genetic base, as well as the existence of programs of prevention of hereditary diseases.

ACADEMIC SKILLS

- Know the basic aspects of human genetics in order to understand describe and interpret the different mechanisms of character transmission in humans.

- Integrate, correlate and coordinate information from different sources and approaches.

- Relate knowledge of genetics with health alterations caused by genetic factors in the field of study of the subject.

- Be able to communicate easily and clearly the concepts of clinical genetics to patients.

LEARNING OUTCOMES

- Value and understand that, for an integral health care, it is fundamental to know the role of Genetics in the different aspects in which health practice is developed: the diagnosis, the treatment and the prevention of human pathologies.
- Be able to describe the structure, function, and transmission mechanisms of genes.
- Identify the patterns of monogenic inheritance and the bases of multifactorial and mitochondrial diseases.
- Describe the techniques of studying chromosomes and the clinical consequences of their anomalies.
- Elaborate and interpret genealogical trees analyzing hereditary transmission mechanisms.
- Apply the knowledge acquired to the resolution and analysis of problems of monogenic and multifactorial inheritance.
- Be able to use and understand the specific terminology of the area of knowledge.
- Effectively use the different information search systems.
- Recognize own limitations and learn to work in a team.

TEACHING ACTIVITIES (theoretical, practical, seminars, workshops, etc.)

THEORETICAL CLASSES

Master lessons. Oral presentation of each topic by the teacher trying to motivate the interest and participation of students in their development.

DIRECTED WORKS
The professor can propose the realization of works whose subject will deepen in aspects of the subject oriented to his application to the Sciences of the Health. These works will be developed in small groups. Students will be able to carry out previously programmed virtual works through the use of computer platforms. These student activities will always be supervised by the teacher.

PRACTICAL CLASSES
The teacher will propose a series of activities, problems and practical cases aimed at the integration and application of the theoretical contents.
The students, always supervised by the teacher, will participate actively in the discussion and resolution of said problems.

OTHER ACTIVITIES
Tutorials. The teacher will assist students to supervise their training, guide them and resolve any doubts that may arise.

CONTENT TOPICS

THEORETICAL CONTENT

Unit 1. Introduction to genetics: gene and genome (nuclear and mitochondrial). Chromatin structure and gene expression.

Unit 2. Human chromosomes. Basic structure. Classification and characteristics of human chromosomes. Cellular division. Origin of chromosomal abnormalities in gametes.

Unit 3. Numerical anomalies of chromosomes. Types. Most frequent aneuploidies: Trisomy of chromosomes 13, 18, 21. Turner syndrome, Klinefelter syndrome.

Unit 4. Structural anomalies of chromosomes. Origin and consequences. Infertility and recurrent miscarriages.

Unit 5. Inheritance patterns. Basic concepts. General patterns of inheritance. Genealogical tree: its importance in the Clinical History. Information and consultation sources.

Unit 6. Autosomal monogenic inheritance. Dominant and recessive inheritance. Autosomal monogenic diseases: Cystic Fibrosis, Muscular Dystrophies, Neuropathies, Connective Diseases.

Unit 7. X-linked inheritance. Diseases with sex-linked inheritance. Color blindness, Hemophilia. Determination of sex and disorders of sexual development.

Unit 8. Factors that complicate the evaluation of inheritance patterns: Incomplete penetrance, variable expressivity: Neurofibromatosis, Tuberous Sclerosis.

Unit 9. Atypical modes of inheritance: Anticipation, imprinting, mitochondrial inheritance. Fragile X Syndrome, Myotonic Dystrophy, Prader Willi and Angelman Syndromes.

Unit 10. Multifactorial inheritance. Genetics of disorders with multifactorial inheritance. Identification of genetic risk factors and influence of environmental factors and lifestyles.

Unit 11. Diagnosis and Prevention of genetic diseases. Prenatal diagnosis. Genetic test. Screening for genetic diseases. Modifiable external factors that interact with the genome.

PRACTICAL CONTENTS

- Meiosis and origin of genetic abnormalities.
- Mitosis. Chromosome identification
- Studying Methods of chromosomes and diagnosis of chromosomal abnormalities.
- Analysis of genealogical trees.
- Analysis and resolution of inheritance problems.
- Knowledge Application of Genetics in the approach of practical cases of diseases.

EVALUATION

The evaluation system is carried out following a continuous evaluation model.

The attendance to the practices will have a value of the 10% of the final grade.

The directed works and/or the resolution of problems/cases proposed in the practices will be evaluated representing the 30% of the total global score.

The 60% of the final grade is obtained through the evaluation of a joint group on the theoretical and practical contents of the subject that refers to the end of the course.

BLIOGRAPHY - INTERNET Resources

- Nussbaum, R.L.; McInnes, R.R.; Willard, H.F.; Thompson & Thompson, Genética en Medicina, Elsevier, 8ª ed., 2016, ISBN 978-84-458-2642-3.

  AVAILABLE IN KLINICALKEY:
  https://www.clinicalkey.com/student/content/toc/3-s2.0-C20150020123

- Turnpenny PD, Ellard S, Ruth Cleaver R, Emery A. Emery's elements of medical genetics and genomics Elsevier 15th Edition. 2021. ISBN: 9780702079665

  AVAILABLE IN KLINICALKEY:
  https://www.clinicalkey.com/student/content/toc/3-s2.0-C20210009603

- Jorde LB, Carey JC, Bamshad MJ, Genética Médica, Elsevier. 6ª Edición. 2020. ISBN: 9788491137979

  AVAILABLE IN KLINICALKEY:
  https://www.clinicalkey.com/student/content/toc/3-s2.0-C20190053186

- I. Ejarque, M. Orera, P. Lapunzina. Genética pare el médico de familia. Ed. Síntesis, 2017. ISBN: 978-84-9171-072-1

INTERNET Resources:

- GeneReviews: http://www.ncbi.nlm.nih.gov/books/NBK1116/
- PubMed: https://www.ncbi.nlm.nih.gov/pubmed/
- OMIM® - Online Mendelian Inheritance in Man®:
  www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM&itool=toolbar
- Human Genome Project Information:
  http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml

TEACHING STAFF * (It should be indicated whether teachers have completed all their teaching dedication or not)

Teacher Responsible (coordinator):
Name: Mª del Carmen Martínez Mora
(cmmora@ucm.es)

Department: Departmental Section of Cell Biology