



Universitat de Lleida

DEGREE CURRICULUM **HUMAN GENOMICS**

Coordination: FERREZUELO MUÑOZ, FRANCISCO

Academic year 2023-24

Subject's general information

Subject name	HUMAN GENOMICS											
Code	100503											
Semester	2D SEMESTER - DEGREE - JUN/SET											
Typology	<table border="1"> <thead> <tr> <th>Degree</th> <th>Course</th> <th>Typology</th> <th>Modality</th> </tr> </thead> <tbody> <tr> <td>Bachelor's Degree in Medicine</td> <td>1</td> <td>COMMON/CORE</td> <td>Only examination</td> </tr> </tbody> </table>				Degree	Course	Typology	Modality	Bachelor's Degree in Medicine	1	COMMON/CORE	Only examination
Degree	Course	Typology	Modality									
Bachelor's Degree in Medicine	1	COMMON/CORE	Only examination									
Course number of credits (ECTS)	6											
Type of activity, credits, and groups	Only examination											
Coordination	FERREZUELO MUÑOZ, FRANCISCO											
Department	false											
Important information on data processing	Consult this link for more information.											
Language	Catalan 75%											
	Spanish 25%											

Teaching staff	E-mail addresses	Credits taught by teacher	Office and hour of attention
FERREZUELO MUÑOZ, FRANCISCO	francisco.ferrezuelo@udl.cat	0	

Subject's extra information

This course is offered the first year of the degree to provide basic knowledge about gene function and transmission in humans

Learning objectives

The goal of this course is to provide the basic knowledge (see below) that is essential for those students who direct their professional career to areas of medicine where it is necessary to understand the molecular foundation of the pathological process. Genes, as the ultimate determinants of cellular function, are also often the cause for the molecular and cellular alterations that define a pathological framework. Many diseases of uncertain or unknown etiology at this time will surely find an explanation at the molecular level, either as a direct result of somatic genetic alterations, or by the greater predisposition of a particular genetic constitution. In any case, gene therapy will soon be a key element of medical practice, and the medical professional must now know its theoretical basis and, in the near future, its practical ones.

Competences

RAM1 To describe the different forms of gene information, expression and regulation

RAM2 To classify the mechanisms of the different patterns of inheritance

RAM3 To Identify those alterations that can be inherited

RAM4 To use some material and basic techniques in a medical science laboratory

Subject contents

Module 1. Structure and complexity of the human genome

- 1.1 Nucleic Acids
- 1.2 DNA condensation. Chromatin and chromosomes
- 1.3 Structure of the gene at the molecular level
- 1.4 Levels of complexity of the human genome

Module 2. Maintenance and integrity of the human genome

- 2.1 Basic mechanisms of DNA replication
- 2.2 Telomere termination and maintenance
- 2.3 Recombination and transposition
- 2.4 Mutations: types and causative agents
- 2.5 Repair mechanisms

Module 3. Gene expression

- 3.1 The flow of genetic information
- 3.2 Transcription
- 3.3 The processing of eukaryotic messenger RNAs: "capping", polyadenylation and "splicing"
- 3.4 The genetic code
- 3.5 Main molecular components in the process of translating messenger RNAs to proteins
- 3.6 Translation

Module 4. Regulation of gene expression

- 4.1 General concepts
- 4.2 Transcriptional regulation
- 4.3 Epigenetics
- 4.4 Posttranscriptional regulation
- 4.5 CRISPR and gene therapy

Module 5. Genetic Analysis

- 5.1. Chromosomes and inheritance: Mitosis and Meiosis
- 5.2. Inheritance patterns in human families
- 5.3. Genetic variability. Mutations
- 5.4 Human genome mapping
- 5.5 Linkage analysis

Module 6. Genetic Pathologies

- 6.1 Chromosomopathies
- 6.2 Hereditary monogenic diseases
- 6.3 Mitochondrial inheritance diseases
- 6.4 Dynamic mutations and other genetical diseases

Module 7. Population genetics

- 7.1 Concept of the Mendelian population. Genotypic and phenotypic frequencies. Hardy-Weinberg equilibrium, deviations and practical applications
- 7.2 Basic concepts of developmental genetics
- 7.3 Basic concepts of cancer genetics
- 7.4 Evolutionary medicine

Module 8. Genetic diagnosis in Medicine

- 8.1. Chromosomal diagnosis (karyotype)
- 8.2. Molecular diagnosis (direct and indirect)
- 8.3. Genetic advice and ethical aspects

Problem Sessions

- Problems / exercises M5 + 6
- Problems / exercises M7
- Problems / exercises M8

Computer classroom sessions

karyotype simulations

Evaluation

One single examination for the whole course. To pass the course 50% of total score is required.

Bibliography

- Griffiths A, et al. (2008), *Genética (9ª Ed) Ed. MacGraw Hill*
- Pierce, B (2006), *Genética, un enfoque conceptual. Editorial Médica panamericana.*
- Nussbaum RL, et al. (2004), *Thompson & Thompson, Genética en Medicina.* Ed Masson
- Solari AJ. (2004). *Genética Humana, fundamentos y aplicaciones en Medicina.* Editorial Médica Panamericana
- Alberts B, et al. (2015), *Molecular Biology of the Cell.* 6th ed. Garland Science
- Alberts B, et al. (2014), *Essential Cell Biology.* Garland Science
- Lewin B, *Molecular Biology (Full Edition) and Genetics.* Jones & Bartlett
- Strachan T, and Read AP (**on-line**). *Human Molecular Genetics 2.* Garland Science
(ncbi.nlm.nih.gov/books/bv.fcgi?rid=hmg)
- Watson JD, et al. (2014), *Molecular Biology of the Gene.* 7th ed Pearson Education Inc.
- Lynn B. Jorde, John C. Carey, MPH and Michael J. Bamshad. *Medical Genetics*, 4th Edition, Mosby Ed 2010