Universitat de Lleida

DEGREE CURRICULUM

HUMAN GENOMICS

Coordination: FERREZUELO MUÑOZ, FRANCISCO

Academic year 2022-23

HUMAN GENOMICS 2022-23

Subject's general information

Subject name	HUMAN GENOMICS
Code	100503
Semester	2D SEMESTER - DEGREE - JUN/SET
Typology	

Degree	Course	Character	Modality
Bachelor's Degree in Medicine	1	COMMON/CORE	Attendance-based

Course number of credits (ECTS)	6
Type of activity, credits, and groups	

Activity type	PRALAB	PRAULA		TEORIA
Number of credits	0.4	0.6	1	4
Number of groups	8	6	3	1

Coordination	FERREZUELO MUÑOZ, FRANCISCO
Department	BASIC MEDICAL SCIENCES
Important information on data processing	Consult this link for more information.
Language	Catalan 75% Spanish 25%

HUMAN GENOMICS 2022-23

Teaching staff	E-mail addresses	Credits taught by teacher	Office and hour of attention
CASALI TABERNET, ANDREU	andreu.casali@udl.cat	4,2	
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FERREZUELO MUÑOZ, FRANCISCO	francisco.ferrezuelo@udl.cat	3	
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LOPEZ ORTEGA, RICARDO ENRIQUE	ricard.lopez@udl.cat	2,2	
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TORRES ROSELL, JORDI	jordi.torres@udl.cat	2	

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.

Objective	**Activities**	**Class attendance**
Acquire basic knowledge about the role of genes at the molecular level and their transmission in human populations	40 one-hour lectures (single grup)	40
Solve basic problems about genetic analysis.	2 sessions - 2h / grup (3 grups) + 2 sessions - 3h / grup (3 grups)	10
Use and interpret a human genome database and design tools for genetic analysis by PCR. Cytogenetic diagnostic and simulations	2 sessions - 2h / grup (Computer room) (6 grups) 1 session - 2h / grup (Computer room) (6 grups)	4 2
Activity at the laboratory: Analysis of a VNTR by PCR	2 sessions - 2h / grup (12 grups)	4
		60

***Hours Student** = Hours of class attendance + hours of homework

Competences

CG7 Understanding and recognizing the normal structure and function of the human body at the different stages of life and at a molecular, cellular, tissue, organic and systems level.

10 Genetic information, expression and regulation

11 Heredity

27 Manipulation of materials and basic laboratory techniques

Subject contents

Module 1. Structure and complexity of the human genome 4h

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 4h

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 6h

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 4h

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 7h

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 6h

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 5h

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 4h

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 4h
Problems / exercises M7 3h
Problems / exercises M8 3h

Computer classroom sessions

PCR and sequencing of the human genome 2h
Ensembl: human genome database 2h
karyotype simulations 2h

Laboratory session

DNA analysis by PCR and electrophoresis 4h

Methodology

A part of the course is developed in sessions of one hour lectures about theoretical concepts. These concepts are reinforced with problem sessions in medium size groups. Laboratory sessions are dedicated to the analysis of a human polymorphism at the molecular level. There will be also some work with online computer tools and access to a human genome database.

Development plan

Activity		Description	Prof	H	Grups
		(Module title or practical activity)		student	
Theory	M1	Structure and complexity of the human genome	JT	4	1
Theory	M2	Maintenance and integrity of the human genome	JT	4	1
Computer		PCR and human genome sequencing	JT	2	6

Theory	M3	Gene expression	FF	6	1
Theory	M4	Gene expression regulation	FF	4	1
Computer		Ensembl: Human genome database	FF	2	6
Theory	M5	Genetic analysis	AC	7	1
Problems		Problems about genetic analysis	AC	4	3
Theory	M6	Genetic pathologies	RL	6	1
Theory	M7	Population genetics	AC	5	1
Problems		Problems about population genetics	AC	3	3
Theory	M8	Genetic diagnostic	RL	4	1
Problems		Problems about genetic diagnostic	AC	3	3
Computer		Karyotype simulations	RL	2	6
Laboratory		DNA analysis by PCR	FF JC ML PM	2	8
Laboratory		DNA analysis and electrophoresis	FF JC ML PM	2	8
				60	

Evaluation

During the course, there will be two written tests consisting of a part of multiple-choice questions on theoretical and practical knowledge and a part of planning and solving exercises / problems. Each part will have a different weight according to the following scheme:

Activity	Points
First test	40
Second test	50

In addition, attendance at the different teaching activities during the course (theoretical classes, computer practices, laboratory, problems) will be assessed with a maximum of 10 points. This attendance will be controlled randomly.

Attendance to the laboratory session is not mandatory, but to get points for the questions on laboratory session at the first test (5 points) one must have attended this activity. However, the students who repeat the course and went to the laboratory session in a previous course do not need to repeat this activity. We will take into account the questions in the test (which you will have to answer again) and the attendance to the activity.

First test

Contents:

- Theory Modules 1-4
- Computer classes 1-2
- Laboratory sessions

Second test

Contents:

- Theory Modules 5-8
- Problem sessions M5-M8
- Computer class 3

The scores (points) obtained in both tests are added to obtain a final score. To pass the course it is necessary to obtain 50 points in the final score. Those who fail to reach this threshold will be given a second chance in September for THE WHOLE COURSE.

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
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- 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