



## 1. General information

Course: HUMAN GENETICS

Type: CORE COURSE

Degree: 331 - UNDERGRADUATE DEGREE PROGRAMME IN MEDICINE

Center: 10 - FACULTY OF MEDICINE OF ALBACETE

Year: 2

Main language: Spanish

Use of additional  
languages:

Web site:

Code: 34313

ECTS credits: 6

Academic year: 2023-24

Group(s): 10

Duration: C2

Second language: English

English Friendly: Y

Bilingual: N

Lecturer: <b>JOSE DANIEL AROCA AGUILAR</b> - Group(s): 10				
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Lecturer: <b>JESÚS JOSÉ FERRE FERNÁNDEZ</b> - Group(s): 10				
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Lecturer: <b>SILVIA LLORENS FOLGADO</b> - Group(s): 10				
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Lecturer: <b>MONICA MUÑOZ LOPEZ</b> - Group(s): 10				
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## 2. Pre-Requisites

Those required to access the Degree.

## 3. Justification in the curriculum, relation to other subjects and to the profession

The subject "Human Genetics" belongs to the Module I (Morphology, Structure and Function of the Human Body) and to the Subject 1.1 (Development, Structure and Function of the Healthy Human Body at the Molecular and Cellular Level) of the Medicine Teaching Plan. It is a mandatory subject, with 6 ECTS and it is taught during the second semester of the second academic year.

The subject "Human Genetics" includes content on the structure and function of genetic material. The causes and inheritance of both monogenic and complex diseases are also studied, as well as the bases for their genetic diagnosis, prevention and treatment. Finally, the principles of evolution are introduced to understand health and disease from a historical perspective.

The contents and resources of each module will be provided through Moodle.

## 4. Degree competences achieved in this course

## Course competences

Code	Description
1.1	Know the cellular structure and function.
1.10	Information, expression and gene regulation.
1.11	Inheritance.
1.17	Manage basic laboratory materials and techniques.
1.2	Biomolecules.
1.8	Cell cycle.
1.9	Cell differentiation and proliferation.

CT01	Mastery of a second foreign language at level B1 of the Common European Framework of Reference for Languages.
CT03	Correct oral and written communication.
G07	Understand and recognise the normal structure and function of the human body, at the molecular, cellular, tissue, organic and systems levels, in the different stages of life and in the two sexes.
G11	Understand and recognise the effects of growth, development and ageing on the individual and his/her social environment.
G36	Be able to formulate hypotheses, collect and critically evaluate information for problem solving, following the scientific method.
G37	Acquire the basic training for the research activity.

## 5. Objectives or Learning Outcomes

### Course learning outcomes

#### Description

Learn how to design and organise the work. Acquire habits of constancy in the study.

Acquisition of oral, written and communication skills.

Know the cellular structure and function. Biomolecules. Metabolism. Metabolic regulation and integration. Know the basic principles of human nutrition. Cellular communication. Excitable membranes. Cell cycle. Cell differentiation and proliferation. Information, expression and gene regulation. Inheritance. Embryonic development and organogenesis. Homeostasis. Adaptation to the environment.

## 6. Units / Contents

**Unit 1: HISTORICAL INTRODUCTION TO HUMAN GENETICS. STRUCTURAL AND FUNCTIONAL ORGANIZATION OF THE HUMAN GENOME.**

**Unit 2: GENETIC VARIATION: MUTATION, RECOMBINATION AND GENETIC POLYMORPHISM.**

**Unit 3: HUMAN CYTOGENETICS.**

**Unit 4: MONOGENIC INHERITANCE.**

**Unit 5: POLYGENIC INHERITANCE AND COMMON DISEASES.**

**Unit 6: IDENTIFICATION OF DISEASE GENES.**

**Unit 7: GENETIC BASIS OF CANCER.**

**Unit 8: MANIPULATION OF GENETIC MATERIAL.**

**Unit 9: GENE THERAPY.**

**Unit 10: TECHNIQUES FOR THE ANALYSIS OF NUCLEIC ACIDS OF EXPERIMENTAL AND CLINICAL INTEREST.**

**Unit 11: FUNDAMENTALS OF CLINICAL GENETICS.**

**Unit 12: INTRODUCTION TO EVOLUTIONARY MEDICINE.**

## 7. Activities, Units/Modules and Methodology

Training Activity	Methodology	Related Competences (only degrees before RD 822/2021)	ECTS	Hours	As	Com	Description
Class Attendance (theory) [ON-SITE]	Lectures	1.1 1.10 1.11 1.17 1.2 G07 G11 G36 G37	1.8	45	Y	N	Teacher seminars are combined with student presentations, resolution of questions, problems, cases and master classes.
Class Attendance (practical) [ON-SITE]	Practical or hands-on activities	1.1 1.10 1.11 1.17 1.2 G07 G11 G36 G37	0.36	9	Y	Y	In these laboratory activities students learn how a human karyotype is made, as well as the foundation of basic mutation analysis techniques and genetic manipulation.
Study and Exam Preparation [OFF-SITE]	Self-study		3.6	90	Y	N	Study of the theoretical and practical contents of the subject, resolution of cases and problems, preparation of work reports.
Final test [ON-SITE]	Assessment tests		0.24	6	Y	Y	
<b>Total:</b>			<b>6</b>	<b>150</b>			
<b>Total credits of in-class work: 2.4</b>			<b>Total class time hours: 60</b>				
<b>Total credits of out of class work: 3.6</b>			<b>Total hours of out of class work: 90</b>				

As: Assessable training activity

Com: Training activity of compulsory overcoming (It will be essential to overcome both continuous and non-continuous assessment).

## 8. Evaluation criteria and Grading System

Evaluation System	Continuous assessment	Non-continuous evaluation*	Description
Laboratory sessions	20.00%	20.00%	The attitude and participation of the student and the results of the activities carried out in the laboratory (5%) and MCQ exams (10%) will be evaluated.
Assessment of active participation	10.00%	0.00%	Answering questions, problem solving and presentation of objectives will be valued.
Final test	70.00%	80.00%	It will consist of a MCQ test corresponding to the theoretical contents of the subject.
<b>Total:</b>	<b>100.00%</b>	<b>100.00%</b>	

According to art. 4 of the UCLM Student Evaluation Regulations, it must be provided to students who cannot regularly attend face-to-face training activities the passing of the subject, having the right (art. 12.2) to be globally graded, in 2 annual calls per subject, an ordinary and an extraordinary one (evaluating 100% of the competences).

**Evaluation criteria for the final exam:****Continuous assessment:**

The ordinary evaluation will be carried out by an accumulative score system. The theory will be assessed by a final exam (70% of the final score).

The practical knowledge will be evaluated by multiple choice question (MCQ) exams (20%). A minimum of 4.0 points in both the theory final and practical exams is required to sum the corresponding points into the final score. Participation will be assessed based on active and constructive presence in class, answering questions, solving problems and exposing objectives (10%). To pass the subject it will be necessary to achieve a minimum score of 5.0 points.

**Non-continuous evaluation:**

\*Chapter III. Art. 4. 2 b) Any student may change to the non-continuous evaluation modality, by the procedure established by the Center, provided that they have not participated during the period of classes.

in evaluable activities that together represent at least 50% of the total evaluation of the subject. If a student has reached that 50% of evaluable activities or if, in any case, the class period has ended, they will be considered in continuous evaluation without the possibility of changing the evaluation modality.

**Specifications for the resit/retake exam:**

Those students who have not passed the subject by the cumulative points system may take the extraordinary call with the same criteria as in the ordinary call.

**Specifications for the second resit / retake exam:**

\*The same criteria will be followed as for the extraordinary call for the previous year, as stated in the corresponding teaching guides (Art. 13.3. Student Assessment Regulations). This call may be used by students who are in the cases indicated in the Student Assessment Regulations that are in force (currently, Art. 13.1).

9. Assignments, course calendar and important dates	
Not related to the syllabus/contents	
Hours	hours
Final test [PRESENCIAL][Assessment tests]	6
Unit 1 (de 12): HISTORICAL INTRODUCTION TO HUMAN GENETICS. STRUCTURAL AND FUNCTIONAL ORGANIZATION OF THE HUMAN GENOME.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	4.5
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 2 (de 12): GENETIC VARIATION: MUTATION, RECOMBINATION AND GENETIC POLYMORPHISM.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	4.5
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 3 (de 12): HUMAN CYTOGENETICS.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	3
Class Attendance (practical) [PRESENCIAL][Practical or hands-on activities]	3
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 4 (de 12): MONOGENIC INHERITANCE.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	4.5
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 5 (de 12): POLYGENIC INHERITANCE AND COMMON DISEASES.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	4.5
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 6 (de 12): IDENTIFICATION OF DISEASE GENES.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	3
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 7 (de 12): GENETIC BASIS OF CANCER.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	3
Class Attendance (practical) [PRESENCIAL][Practical or hands-on activities]	3
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 8 (de 12): MANIPULATION OF GENETIC MATERIAL.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	3
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 9 (de 12): GENE THERAPY.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	3
Class Attendance (practical) [PRESENCIAL][Practical or hands-on activities]	3
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 10 (de 12): TECHNIQUES FOR THE ANALYSIS OF NUCLEIC ACIDS OF EXPERIMENTAL AND CLINICAL INTEREST.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	4.5
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Unit 11 (de 12): FUNDAMENTALS OF CLINICAL GENETICS.	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	4.5
Study and Exam Preparation [AUTÓNOMA][Self-study]	7.5
Global activity	

Activities	hours
Class Attendance (theory) [PRESENCIAL][Lectures]	42
Class Attendance (practical) [PRESENCIAL][Practical or hands-on activities]	9
Study and Exam Preparation [AUTÓNOMA][Self-study]	82.5
Final test [PRESENCIAL][Assessment tests]	6
<b>Total horas: 139.5</b>	

10. Bibliography and Sources						
Author(s)	Title/Link	Publishing house	Citv	ISBN	Year	Description
Jorde, Carey & Bamshad	Genética médica	Elsevier		9788491137979	2020	
Randolph M. Nesse & George C. Williams	¿Por qué enfermamos?	Grijalbo		9788425334580	2000	
Tom Strachan, Andrew Read	Human molecular genetics	CRC Press		9780815345893	2019	
Peter Turnpenny Sian Ellard Ruth Cleaver	Emery's Elements of Medical Genetics and Genomics 16th Edition	Elsevier		9780702079665	2021	
Nussbaum, McInnes & Willard	Thompson & Thompson. Genetica en medicina	Elsevier		9788445826423	2016	