



## 1. General information

Course: HUMAN GENETICS

Type: CORE COURSE

Degree: 332 - UNDERGRADUATE DEGREE PROGRAMME IN MEDICINE

Center: 9 - FACULTY OF MEDICINE OF CIUDAD REAL

Year: 2

Main language: Spanish

Use of additional languages: English

Web site: [http://www.uclm.es/cr/medicina/grado\\_plan\\_estudios.html](http://www.uclm.es/cr/medicina/grado_plan_estudios.html)

Code: 34313

ECTS credits: 6

Academic year: 2023-24

Group(s): 20

Duration: C2

Second language:

English Friendly: Y

Bilingual: N

Lecturer: INMACULADA BALLESTEROS YAÑEZ - Group(s): 20				
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## 2. Pre-Requisites

In accordance with the prerequisites and incompatibilities established in the curriculum of the Degree in Medicine of the University of Castilla-La Mancha, in order to pass the subject of Human Genetics students must have previously passed the subjects of Biology and Biochemistry I of the first year.

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

The Study Plan of the Degree in Medicine is adapted to the requirements established in the Royal Decree 1393/2007, of October 29th (B. O. E. of October 30th, 2007), in the Order ECI/332/2008, of February 13th (B. O. E. of February 15th, 2008) and in the agreements adopted by the Medicine Degree Commission of May 28th, 2008 and June 30th, 2008. The degree of Graduate in Medicine consists, according to current legislation, of 360 ECTS distributed over 6 academic years and structured in at least 5 modules, with defined competences, as the medical profession is a regulated profession. The five modules of common subjects are defined by the content of the subjects that include compulsory subjects for the Degree in Medicine. The sixth module has been defined according to the optional nature of the subjects that comprise it. In the first two years of the degree, the necessary basic preclinical subjects are taught (Modules 1 and 2) that will provide students with the necessary elements for the foundation of the clinical knowledge and medical skills that they will acquire later (Modules 3, 4, 5 and 6), promoting the early insertion of students in health centers. The subject of Human Genetics is part of Module 1 ("Morphology, Structure and Function of the Human Body"), within the subject (M1.1) entitled "Development, Structure and Function of the Healthy Human Body at Molecular and Cellular Level", being a Compulsory Training Subject. The learning of Human Genetics is established around theoretical and practical contents, oriented to the acquisition of the competences defined in Module 1, according to the Memory approved by ANECA for the Degree of Medicine by this University. Human Genetics plays an essential role in the medical training of the XXI century, since it is a discipline in which important aspects of different clinical specialties are integrated. It is subject to continuous conceptual and technological advances. The general objective of this course is for students to acquire an adequate knowledge of the structure and function of hereditary material, to understand the laws that govern the transmission of normal and pathological biological characters, and to know the main diagnostic and therapeutic applications currently offered by human genetics.

## 4. Degree competences achieved in this course

Course competences	
Code	Description
1.1	Knowledge of cell structure and function.
1.10	Information, expression and gene regulation.
1.11	Inheritance.
1.13	To know the morphology, structure and function of the skin, blood, circulatory, digestive, locomotor, reproductive, excretory and respiratory apparatus and systems; endocrine system, immune system and central and peripheral nervous system.
1.15	Homeostasis
1.17	Handling basic laboratory material and techniques.
1.18	Interpret a normal blood test.
1.19	Recognize with macroscopic and microscopic methods and imaging techniques the morphology and structure of tissues, organs and systems.
1.2	Biomolecules.
1.3	Metabolism.
1.4	Metabolic regulation and integration.
1.5	To know the basic principles of human nutrition.

1.6	Cellular communication.
1.7	Excitable membranes.
1.8	Cell cycle.
1.9	Cell differentiation and proliferation.
CT01	Proficiency in a second foreign language at level B1 of the Common European Framework of Reference for Languages.
CT03	Good oral and written communication skills.
G07	Understand and recognize the normal structure and function of the human body, at the molecular, cellular, tissue, organic and system levels, in the different stages of life and in both sexes.
G11	Understand and recognize the effects of growth, development and aging on the individual and their social environment.
G36	To be able to formulate hypotheses, collect and critically evaluate information for problem solving, following the scientific method.
G37	To acquire the basic training for research activity.

## 5. Objectives or Learning Outcomes

### Course learning outcomes

#### Description

Learning to design and organize the work. Acquiring habits of perseverance in the study.

Acquisition of oral and/or written presentation and communication skills.

To know the cellular structure and function. Biomolecules. Metabolism. Metabolic regulation and integration. To know the basic principles of human nutrition.

Cell communication. Excitable membranes. Cell cycle. Cell differentiation and proliferation. Gene information, expression and regulation. Inheritance. Embryonic development and organogenesis. Homeostasis. Adaptation to the environment.

For the subject of Human Genetics additional outcomes are planned: To know the structure and function of the human genome. To know the principles of heredity. To understand the genetic basis of normal and pathological traits. To know the diagnostic and therapeutic applications of genetics.

Handling basic laboratory material and techniques. Interpret a normal blood test. Recognize with macroscopic and microscopic methods and imaging techniques the morphology and structure of tissues. Perform functional tests, determine vital parameters and interpret them. Basic physical examination.

### Additional outcomes

To know the concepts of genetics and heredity, gene and genome. Gene expression and regulation. To know the structure and function of the human genome.

To know the structure of human chromosomes. To know the concepts of mutation and genetic variation. To know the genetic basis of heredity and monogenic and complex human diseases. To know the techniques of genetic and genomic analysis. To know the chromosomal, genomic and molecular bases of genetic diseases. To know the genetics of cancer. To know the basics of clinical genetics, genetic diagnosis and genetic counseling. Genetics as therapy and treatment of genetic disease.

## 6. Units / Contents

### Unit 1: INTRODUCTION TO GENETICS. GENES, GENOME AND HUMAN CHROMOSOMES

**Unit 1.1** Introduction to Genetics in Medicine.

**Unit 1.2** Structure, organization and architecture of the human genome.

**Unit 1.3** Epigenetics.

**Unit 1.4** Genetic variation.

### Unit 2: INHERITANCE

**Unit 2.1** Mendelian inheritance.

**Unit 2.2** Non-Mendelian monogenic inheritance.

**Unit 2.3** Inheritance of common diseases.

**Unit 2.4** Epigenetic factors in inheritance and disease.

**Unit 2.5** Genetic maps and gene identification.

### Unit 3: GENOMIC, CHROMOSOMAL AND MOLECULAR BASIS OF GENETIC DISEASE

**Unit 3.1** Cytogenetics and chromosomal syndromes.

**Unit 3.2** Genomic disorders. Microdeletion and microduplication syndromes.

**Unit 3.3** Cancer genetics.

### Unit 4: GENETIC AND MOLECULAR ANALYSIS TECHNIQUES. GENETIC ENGINEERING

**Unit 4.1** Genetic analysis techniques.

**Unit 4.2** Principles of genetic engineering.

**Unit 4.3** Principles of gene therapy.

### Unit 5: CLINICAL GENETICS AND EVOLUTIONARY MEDICINE

**Unit 5.1** Application of genetic medicine and genetic analysis in the clinic.

**Unit 5.2** Precision treatment of genetic disease.

**Unit 5.3** Principles of 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 1.13 1.15 1.17 1.18 1.19 1.2 1.3 1.4 1.5 1.6 1.7 1.8 1.9 CT01 CT03 G07 G11 G36 G37	1.4	35	N	-	1, 2 Phases
Class Attendance (practical) [ON-SITE]	Practical or hands-on activities	1.1 1.10 1.11 1.13 1.15 1.17 1.18 1.19 1.2 1.3 1.4 1.5 1.6 1.7 1.8 1.9 CT01 CT03 G07 G11 G36 G37	0.36	9	Y	Y	Laboratory and bioinformatics practicals. This part is mandatory.
Study and Exam Preparation [OFF-SITE]	Self-study	1.1 1.10 1.11 1.13 1.15 1.17 1.18 1.19 1.2 1.3 1.4 1.5 1.6 1.7 1.8 1.9 CT01 CT03 G07 G11 G36 G37	3.4	85	N	-	

Progress test [ON-SITE]	Assessment tests	1.1 1.10 1.11 1.13 1.15 1.17 1.18 1.19 1.2 1.3 1.4 1.5 1.6 1.7 1.8 1.9 CT01 CT03 G07 G11 G36 G37	0.16	4	Y	N	5 Phase
Final test [ON-SITE]	Assessment tests	1.1 1.10 1.11 1.13 1.15 1.17 1.18 1.19 1.2 1.3 1.4 1.5 1.6 1.7 1.8 1.9 CT01 CT03 G07 G11 G36 G37	0.08	2	Y	N	Final exam (ordinary call)
Project or Topic Presentations [ON-SITE]	Guided or supervised work	1.1 1.10 1.11 1.13 1.15 1.17 1.18 1.19 1.2 1.3 1.4 1.5 1.6 1.7 1.8 1.9 CT01 CT03 G07 G11 G36 G37	0.6	15	Y	N	Presentation of clinical cases, problem solving (Phase 3-4). Optional assignments.
Total:			6	150			
Total credits of in-class work: 2.6			Total class time hours: 65				
Total credits of out of class work: 3.4			Total hours of out of class work: 85				

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
Progress Tests	50.00%	0.00%	Module exams (5 exams with a weight of 10% each). To pass the course it will be necessary to obtain in the theoretical part at least 40% of the 70% and to fulfill the requirements of the section "Evaluation criteria" of the electronic guide.
Final test	20.00%	70.00%	Final exam (ordinary or extraordinary call). In order to pass the course, it will be necessary to obtain at least 40% of the 70% in the theoretical part and to fulfill the requirements of the section "Evaluation criteria" of the electronic guide.
Laboratory sessions	25.00%	25.00%	Bioinformatics and laboratory practicals and practical exam. The completion of the practicals is MANDATORY to pass the course. In addition, to pass the course it will be necessary to obtain in the practical part at least 40% of the 30% and to fulfill the requirements of the section "Evaluation criteria" of the electronic guide.
Assessment of active participation	5.00%	5.00%	Class participation and attitude. Presentations. Voluntary work. To pass the course it will be necessary to obtain in the practical part at least 40% of the 30% and to fulfill the requirements of the section "Evaluation criteria" of the electronic guide.
<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:

A student enrolled for the first time in a subject is entitled to two calls during the academic year:

1. Ordinary call: it comprises the continuous evaluation of all theoretical and practical activities reflected in the timetable attending to the conditions described in the teaching guide of the subject and the minimum attendance requirements to pass the subject.
2. Extraordinary call: It includes the evaluation exclusively of the failed part of the subject in the ordinary call. It consists of a theoretical exam and/or a practical exam. The rest of the marks of the practical part will be those obtained during the course in reports, seminars, presentations, assignments, participation and attitude or OSCE.

In case of failing the course the first time it is taken, for the following academic year there will be two of this three options:

1. Ordinary call: within this call, two modalities can be chosen:

a. Attendance mode: It includes the continuous evaluation of all theoretical and practical activities reflected in the timetable, complying with the conditions described in the teaching guide of the subject, as if the subject was taken for the first time. Thus, the grades obtained in the previous year will not be taken into account.

b. Non-attendance mode: It includes the evaluation of only the failed part of the subject in the previous course through a theoretical exam and/or a practical exam per semester on the same date as the final exam of each semester. The marks for practical exams other than the practical exam will be kept from the previous course. This modality can only be chosen in the case of having taken the subject in the ordinary call in the previous academic year.

2. Extraordinary call: It includes the evaluation of only the failed part of the subject in the ordinary call either of the current academic year, if the student has chosen the ordinary on-site call, or of the previous academic year, in the rest of the cases. It will consist of a theoretical and/or a practical exam. The rest of the marks of the practical part will be those of the current or previous course. In the case of not having taken the ordinary on-site exam in the current or previous academic year, the grades of previous exams will not be taken into account, since only one course will be kept.

3. Special final exam: This includes the evaluation of only the failed part of the subject in the previous academic year. This call can only be requested in key subjects. It will consist of a theoretical exam and/or a practical exam. The rest of the marks of the practical part will be those of the previous course. In the

case of not having taken the ordinary on-site exam in the current or previous course, the grades of previous exams will not be taken into account since only one course will be kept.

These conditions will only be maintained in the academic year consecutive to the ordinary on-site call of a subject. The grade of the practical or theoretical part passed will only be kept if the minimum attendance requirements to pass the subject described in the electronic guide have been met.

If the subject is not passed in the second academic year, the same biannual cycle criteria described for the first and second year of enrollment will be followed in the third and successive odd numbered years of enrollment.

#### ORDINARY CALL:

Theoretical evaluation:

70% distributed in:

- 50% module exams
- 20% final semester exams

To pass the course it will be necessary to obtain 40% of this 70%, which means, at least, 2.8 points in the theoretical part of the 10 total points of the course and to fulfill the requirements of the section "Evaluation criteria".

For the module exams (50% of the grade, i.e. 5 points) no minimum grade is established, so all grades obtained will be added together.

For the final exams (20% of the grade, i.e. 2 points) a minimum grade equal to 40% of the maximum grade to be achieved in each final exam is established.

In order to favor the weight of the continuous evaluation in the final grade, not reaching the minimum grade established will not mean the impossibility to pass the course, but the points of that final exam will not be added to the rest of the points obtained.

Evaluation of practices, presentations, problems, assignments, participation and attitude:

30% valued jointly as follows for basic subjects:

- Practical exams at the end of the semester: 25%.
- Class participation, presentations, papers and attitude: 5%.

30% evaluated jointly as follows for clinical subjects from 3rd to 5th year:

- OSCE (Objective Structured Clinical Evaluation): 15%
- Practical exam: 10%.
- Participation and attitude: 5%.

To pass the course it will be necessary to obtain 40% of the 30%, which represents at least 1.2 points in the practical part of the 10 total points of the course and to fulfill the requirements of the section "Evaluation Criteria" that appears in the electronic guide.

The indispensable requirement for the student to be able to take the OSCE test in a course is to be enrolled in at least 75% of the subjects of that course that participate in this OSCE. test. Otherwise, an alternative practical test will be given independently for each subject.

#### EXTRAORDINARY CALL, SPECIAL FINAL CALL, ORDINARY NON-ATTENDANCE CALL:

Theoretical evaluation: exam with a weight of 70%. To pass the course it will be necessary to obtain 40% of this 70%, which means at least 2.8 points in the theoretical part of the 10 total points of the course and meet the requirements of the section "Evaluation criteria". In case of having passed the theoretical part of the course in the current or previous year, the grade obtained in the last exam will be maintained.

Practical evaluation: to pass the course it will be necessary to obtain 40% of the 30%, which means at least 1.2 points in the practical part of the 10 total points of the course and to fulfill the requirements of the section "Evaluation criteria". In case of having passed the practical part in the current or previous course, the grade obtained in the last exam will be maintained.

There will be a Practice Exam and the results obtained in the OSCE of the previous ordinary on-site exam, either of the current or previous year, will be taken into account.

#### Non-continuous evaluation:

See what is described in the previous point.

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

See what is described in the previous point.

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

See what is described in the previous point.

9. Assignments, course calendar and important dates	
Not related to the syllabus/contents	
<b>Hours</b>	<b>hours</b>
Class Attendance (practical) [PRESENCIAL][Practical or hands-on activities]	9
Study and Exam Preparation [AUTÓNOMA][Self-study]	85
Progress test [PRESENCIAL][Assessment tests]	4
Final test [PRESENCIAL][Assessment tests]	2
Project or Topic Presentations [PRESENCIAL][Guided or supervised work]	15
Unit 1 (de 5): INTRODUCTION TO GENETICS. GENES, GENOME AND HUMAN CHROMOSOMES	
<b>Activities</b>	<b>Hours</b>
Class Attendance (theory) [PRESENCIAL][Lectures]	7
Group 20:	
<b>Initial date:</b> 29-01-2024	<b>End date:</b> 16-02-2024
Unit 2 (de 5): INHERITANCE	
<b>Activities</b>	<b>Hours</b>
Class Attendance (theory) [PRESENCIAL][Lectures]	7
Group 20:	
<b>Initial date:</b> 19-02-2024	<b>End date:</b> 08-03-2024
Unit 3 (de 5): GENOMIC, CHROMOSOMAL AND MOLECULAR BASIS OF GENETIC DISEASE	
<b>Activities</b>	<b>Hours</b>
Class Attendance (theory) [PRESENCIAL][Lectures]	7
Group 20:	
<b>Initial date:</b> 11-03-2024	<b>End date:</b> 05-04-2024

Unit 4 (de 5): GENETIC AND MOLECULAR ANALYSIS TECHNIQUES. GENETIC ENGINEERING	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	7
Group 20:	
Initial date: 08-04-2024	End date: 26-04-2024
Unit 5 (de 5): CLINICAL GENETICS AND EVOLUTIONARY MEDICINE	
Activities	Hours
Class Attendance (theory) [PRESENCIAL][Lectures]	7
Group 20:	
Initial date: 29-04-2024	End date: 17-05-2024
Global activity	
Activities	hours
Class Attendance (theory) [PRESENCIAL][Lectures]	35
Class Attendance (practical) [PRESENCIAL][Practical or hands-on activities]	9
Study and Exam Preparation [AUTÓNOMA][Self-study]	85
Progress test [PRESENCIAL][Assessment tests]	4
Final test [PRESENCIAL][Assessment tests]	2
Project or Topic Presentations [PRESENCIAL][Guided or supervised work]	15
Total horas: 150	

10. Bibliography and Sources						
Author(s)	Title/Link	Publishing house	Citv	ISBN	Year	Description
Nussbaum, McInnes, Willard	Genetics in Medicine	Elsevier		978-1-4377-0696-3	2016	Octava edición
Tom Strachan	Genetics and Genomics in Medicine	Garland science		978-0-8153-4480-3	2015	
Tom Strachan	Human Molecular Genetics	Garland science		978-0-8153-4149-9	2010	
Jorde, Carey, Bamshad	Genética Médica	Elsevier		978-84-9113-797-9	2020	Sexta edición
Lewis R.	Human Genetics: concepts and aplicaciones	Mc Graw Hill		978-1-259-09563-4	2015	
Peter D. Turnpenny, Sian Ellard, Ruth Cleaver	Elementos de Genética Médica y Genómica	Elsevier		978-84-1382-168-9	2022	16ª Edición