Information about the course

Degree: Bachelor of Science Degree in Medicine

Faculty: Faculty of Medicine and Health Sciences

Code: 342002 **Name:** Genetics

Credits: 3,00 ECTS **Year:** 1 **Semester:** 1

Module: Diagnostic and therapeutical procedures.

Subject Matter: Procedimientos diagnósticos **Type:** Optativa

Branch of knowledge:

Department: Pathology

Type of learning: Classroom-based learning

Language/-s in which it is given: Spanish

Teachers:

341A	Francisco Jose Revert Ros **(Profesor responsable)**	fj.revert@ucv.es
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Module organization

Diagnostic and therapeutical procedures.

Subject Matter	ECTS	Subject	ECTS	Year/semester
Procedimientos diagnósticos	39	Basic Immunology	3	1/2
		Functional Assessment	6	1/2
		Genetics	3	1/1
		Introduction to Medicine	3	1/2
		Laboratory of Diagnostic Tests	3	5/1
		Medical Microbiology and Parasitology	6	3/1
		Pathological Anatomy	6	2/2
		Physiological Records and Functional Tests	3	2/2
		Radiodiagnostic and Imaging Techniques	6	3/1
Procedimientos terapéuticos	27	Anaesthesia and Resuscitation	3	5/1
		Biotechnology	6	1/2
		General and Special Pharmacology	9	3/2
		General Procedures of Intervention	6	1/2
		Rehabilitation and Physical Therapy	3	4/2

Recommended knowledge

Knowledge of High School Biology.

Learning outcomes

Al finalizar la asignatura, el estudiante deberá demostrar haber adquirido los siguientes resultados de aprendizaje:

R1 - Know the techniques used in Molecular Genetics.

Learning outcomes of the specified title

Type of AR: Description

- Assessing the risk-benefit ratio of diagnostic and therapeutic procedures

- Knowing how to use information and communication technologies in clinical, therapeutic, preventive and research activities

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Type of AR: Description

- Knowing the characteristics of tissues in different situations of injury, adaptation and cell death. Inflammation. Cell growth disturbances. Pathological anatomy of the different devices and systems. Biochemical, cytogenetic and molecular biology markers applied to clinical diagnosis

- Knowing the indications of biochemical, haematological, immunological, microbiological, anatomopathological and imaging tests

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Type of AR: Description

- Basic knowledge of the National Health System and health legislation

- Developing professional practice with respect for other health professionals, acquiring teamwork skills

- Developing professional practice with respect to patient autonomy, beliefs and culture

- Knowing how to apply the principle of social justice to professional practice and understanding the ethical implications of health in a changing global context

- Maintaining and using records with patient information for further analysis, preserving data confidentiality

- Recognizing the essential elements of the medical profession, including ethical principles, legal responsibilities, and patient-centered professional exercise

- Recognizing the limitations themselves and the need to maintain and update their professional competence, giving special importance to the autonomous learning of new knowledge and techniques and to the motivation for quality

- Students can pass on information, ideas, problems and solutions to both a specialized and non-specialized audience

- Students have demonstrated to possess and understand knowledge in a study area that starts from the base of the general secondary education, and is usually found at a level that, while supported by advanced textbooks, also includes some aspects that involve knowledge from the forefront of their field of study

- Students have developed the learning skills needed to undertake further studies with a high degree of autonomy

- Students have the ability to collect and interpret relevant data (usually within their area of study) to make judgments that include a reflection on relevant social, scientific or ethical topics

- Students know how to apply their knowledge to their job or vocation in a professional way and possess the competences that are usually demonstrated through the elaboration and defense of arguments and the resolution of problems within their area of study

- Understanding the importance of such principles for the benefit of the patient, society and profession, with special attention to professional secrecy

R2 - Know the fundamental characteristics of the human genome and current genome sequencing methods.

Learning outcomes of the specified title

Type of AR: Description

- Assessing the risk-benefit ratio of diagnostic and therapeutic procedures

- Knowing how to use information and communication technologies in clinical, therapeutic, preventive and research activities

Type of AR: Description

- Knowing the characteristics of tissues in different situations of injury, adaptation and cell death. Inflammation. Cell growth disturbances. Pathological anatomy of the different devices and systems. Biochemical, cytogenetic and molecular biology markers applied to clinical diagnosis

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R3 - Know the concepts around genetic variation.

Learning outcomes of the specified title

Type of AR: Description

- Assessing the risk-benefit ratio of diagnostic and therapeutic procedures

- Knowing how to use information and communication technologies in clinical, therapeutic, preventive and research activities

Type of AR: Description

- Knowing the characteristics of tissues in different situations of injury, adaptation and cell death. Inflammation. Cell growth disturbances. Pathological anatomy of the different devices and systems. Biochemical, cytogenetic and molecular biology markers applied to clinical diagnosis

- Knowing the indications of biochemical, haematological, immunological, microbiological, anatomopathological and imaging tests

Type of AR: Description

- Basic knowledge of the National Health System and health legislation

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R4 - Distinguish the different chromosomal abnormalities that may occur in humans.

Learning outcomes of the specified title

Type of AR: Description

- Assessing the risk-benefit ratio of diagnostic and therapeutic procedures

- Knowing how to use information and communication technologies in clinical, therapeutic, preventive and research activities

Type of AR: Description

- Knowing the characteristics of tissues in different situations of injury, adaptation and cell death. Inflammation. Cell growth disturbances. Pathological anatomy of the different devices and systems. Biochemical, cytogenetic and molecular biology markers applied to clinical diagnosis

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Type of AR: Description

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R5 - Differentiate the different types of genetic alterations that cause disease.

Learning outcomes of the specified title

Type of AR: Description

- Assessing the risk-benefit ratio of diagnostic and therapeutic procedures

- Knowing how to use information and communication technologies in clinical, therapeutic, preventive and research activities

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Type of AR: Description

- Knowing the characteristics of tissues in different situations of injury, adaptation and cell death. Inflammation. Cell growth disturbances. Pathological anatomy of the different devices and systems. Biochemical, cytogenetic and molecular biology markers applied to clinical diagnosis

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R6 - Know the basics for the diagnosis of genetic diseases and is able to discern the right one for each case.

Learning outcomes of the specified title

Type of AR: Description

- Assessing the risk-benefit ratio of diagnostic and therapeutic procedures

- Knowing how to use information and communication technologies in clinical, therapeutic, preventive and research activities

Type of AR: Description

- Knowing the characteristics of tissues in different situations of injury, adaptation and cell death. Inflammation. Cell growth disturbances. Pathological anatomy of the different devices and systems. Biochemical, cytogenetic and molecular biology markers applied to clinical diagnosis

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Assessment system

Modalidad presencial

Assessed learning outcomes	Granted percentage	Assessment tool
	70,00%	Tests
	30,00%	Practices

Observations

The evaluation will have the following sections:
- Final exam of the course (multiple choice): 6 points.
- Quizzes (multiple choice): 1 point total.
- Lab report: 1 point
- Practical exercises: 2 points

The maximum overall grade is 10, and at least a 5-grade must be achieved to pass the course.

The overall grade of the course will be calculated taking into account all the above mentioned evaluable sections, as long as at least 50% is obtained in the final exam, which will include both theory and practical questions (laboratory and practical exercises).

CRITERIA FOR THE AWARDING OF HONORS:
According to Article 22 of the Regulatory Regulations for the Evaluation and Grading of UCV Subjects, Honor Mentions (HM) may be awarded to students who have obtained an A grade. The number of HM awarded may not exceed five percent of the students included in the same official transcript, unless this is less than 20, in which case only one HM will be awarded.

MENTION OF DISTINCTION:
In accordance with the regulations governing the assessment and grading of subjects in force at UCV, the distinction of "Matrícula de Honor" (Honours with Distinction) may be awarded to students who have achieved a grade of 9.0 or higher. The number of "Matrículas de Honor" (Honours with Distinction) may not exceed five percent of the students enrolled in the group for the corresponding academic year, unless the number of enrolled students is fewer than 20, in which case a single "Matrícula de Honor" (Honours with Distinction) may be awarded. Exceptionally, these distinctions may be assigned globally across different groups of the same subject. Nevertheless, the total number of distinctions awarded will be the same as if they were assigned by group, but they may be distributed among all students based on a common criterion, regardless of the group to which they belong. The criteria for awarding "Matrícula de Honor" (Honours with Distinction) will be determined according to the guidelines stipulated by the professor responsible for the course, as detailed in the "Observations" section of the evaluation system in the course guide.

Actividades formativas

The methodologies to be used so that the students reach the expected learning outcomes will be the following:

M1	Masterclass
M2	Problems resolution and practical cases
M4	Content presentations by teacher
M5	Knowledges and skills explanation
M6	Laboratory practices
M9	Knowledge acquirance through student interaction and activity
M11	Personalised attention by professor
M12	Tests to understand the level of knowledge acquirance and skills
M14	Online activity on e-learning
M15	Personal study

M16 Information research

M18 Work in team

IN-CLASS TRAINING ACTIVITIES

ACTVITY	RELATIONSHIP WITH THE COURSE LEARNING OUTCOMES	METHODOLOGY	HOURS	ECTS
Theory class	R1, R2, R3, R4, R5, R6	Masterclass Problems resolution and practical cases Content presentations by teacher Knowledges and skills explanation Knowledge acquirance through student interaction and activity	22,00	0,88
Seminar and group practices	R1, R2, R3, R4, R5, R6	Knowledges and skills explanation Laboratory practices Personalised attention by professor	4,00	0,16
Tutoring	R1, R2, R3, R4, R5, R6	Personalised attention by professor	2,00	0,08
Evaluation	R1, R2, R3, R4, R5, R6	Tests to understand the level of knowledge acquirance and skills Online activity on e-learning	2,00	0,08

TOTAL			**30,00**	**1,20**

TRAINING ACTIVITIES OF AUTONOMOUS WORK

ACTVITY	RELATIONSHIP WITH THE COURSE LEARNING OUTCOMES	METHODOLOGY	HOURS	ECTS
No attendance	R1, R2, R3, R4, R5, R6	Problems resolution and practical cases Online activity on e-learning Personal study Information research Work in team	45,00	1,80
TOTAL			**45,00**	**1,80**

Description of contents

Descripción de contenidos necesarios para la adquisición de los resultados de aprendizaje.

Theoretical content:

Block of content	Contents
SECTION I: THE HUMAN GENOME	**Chapter 1:** Genetics and genomics in Medicine. **Chapter 2:** Introduction to the genome human. ·The human genome and the chromosomal bases of inheritance. ·Variation in the human genome. ·Transmission of the genome. ·Human gametogenesis and fertilization. ·Medical importance of mitosis and meiosis. **Chapter 3:** Structure and function of human genes. ·The central dogma: DNA -> RNA -> protein. ·Gene expression. ·Variation of gene expression and its importance in medicine.

SECTION II: ALTERATION OF THE HUMAN GENOME	**Chapter 4:** Human genetic diversity: mutation and polymorphism. ·Origin and frequency of different types of mutations. ·Types of mutations and their consequences. ·Variation in individual genomes. **Chapter 5:** Clinical cytogenetics and genomic analysis. ·Chromosome analysis. ·Genomic analysis. ·Chromosomal abnormalities. **Chapter 6:** Chromosomal and genomic bases of the disease: disorders of the autosomes and sex chromosomes. ·Mechanisms of anomalies. ·Aneuploidy. ·Uniparental disomy. ·Microdeletion and duplication syndromes. ·Disorders associated with genomic imprinting. ·Sex chromosomes and their abnormalities.
SECTION III: PATTERNS OF INHERITANCE	**Chapter 7:** Monogenic inheritance. ·Family trees. ·Autosomal Mendelian inheritance. ·X-linked inheritance. ·Mosaicism. ·Dynamic mutations: expansion of unstable repeats. ·Inheritance of disorders caused by mutations in the mitochondrial genome. **Chapter 8:** Complex inheritance. ·Qualitative and quantitative features. ·Examples of common multifactorial diseases with genetic contribution. ·Examples of multifactorial traits with known specific genetic and environmental factors.

BLOCK IV: GENETIC AND MOLECULAR BASES OF HUMAN GENETIC DISEASES	**Chapter 9:** Identification of the genetic bases of human diseases. ·Genetic basis for linkage analysis and association. ·Mapping of human genes causing diseases. ·From gene mapping to gene identification. ·Searching for genes by sequencing the genome **Chapter 10:** Molecular bases of the genetic diseases. ·How mutations affect synthesis and function of proteins. ·Relationship between genotype and phenotype.
SECTION V: GENETICS AND GENOMICS OF CANCER	**Chapter 11:** Genetics and genomics of cancer. ·Genetic bases of cancer. ·Familial and sporadic cancer. ·Cancer and the environment.
SECTION VI: CLINICAL GENETICS	**Chapter 12:** Risk assessment and genetic counseling. **Chapter 13:** Prenatal diagnosis. **Chapter 14**: Ethical aspects in genetics and genomics.

Temporary organization of learning:

Block of content	Sessions	Hours
SECTION I: THE HUMAN GENOME	3	6,00
SECTION II: ALTERATION OF THE HUMAN GENOME	3	6,00
SECTION III: PATTERNS OF INHERITANCE	4	8,00
BLOCK IV: GENETIC AND MOLECULAR BASES OF HUMAN GENETIC DISEASES	3	6,00
SECTION V: GENETICS AND GENOMICS OF CANCER	1	2,00
SECTION VI: CLINICAL GENETICS	1	2,00

References

EMERY. Elementos de genética médica y genómica, 16.ª Edición 2022 Elsevier España, S.L.U.

JORDE, L.B., CAREY, J.C., BAMSHAD, M.J., Genética Médica, 6 Ed, Elsevier, 2020.

NUSSBAUM, R.L., McINNES, R.R., WILLARD, H.F., Thompson y Thompson. Genética en Medicina, 8 Ed, Elsevier, 2016.

Bibliografía complementaria: NOVO, FJ., Genética Humana: conceptos, mecanismos y aplicaciones de la genética en elcampo de la Biomedicina, Pearson Prentice Hall, 2007.