



Information about the subject

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: Diagnostic procedures **Type:** Elective

Field of knowledge: Health Science

Department: -

Type of learning: Classroom-based learning

Languages in which it is taught: Spanish

Lecturer/-s:

341A Francisco Jose Revert Ros (**Responsible Lecturer**) fj.revert@ucv.es

341B Francisco Jose Revert Ros (**Responsible Lecturer**) fj.revert@ucv.es



Module organization

Diagnostic and therapeutical procedures.

Subject Matter	ECTS	Subject	ECTS	Year/semester
Diagnostic procedures	39,00	Basic Immunology	3,00	1/2
		Functional Assessment	6,00	This elective is not offered in the academic year 23/24
		Genetics	3,00	1/1
		Introduction to Medicine	3,00	1/2
		Laboratory of Diagnostic Tests	3,00	5/1
		Medical Microbiology and Parasitology	6,00	3/1
		Pathological Anatomy	6,00	2/2
		Physiological Records and Functional Tests	3,00	2/2
		Radiodiagnostic and Imaging Techniques	6,00	3/1
Therapeutic procedure	27,00	Anaesthesia and Resuscitation	3,00	5/1
		Biotechnology	6,00	This elective is not offered in the academic year 23/24
		General and Special Pharmacology	9,00	3/2



Therapeutic
procedure

General Procedures of
Intervention

6,00

This elective is not
offered in the
academic year
23/24

Rehabilitation and
Physical Therapy

3,00

4/2

Recommended knowledge

It is recommended to know the structure of cells and specifically the organization of DNA in the cell nucleus, as well as its main characteristics:

- DNA as a genetic substance.
- Structure of nucleic acids.
- Physical-chemical properties of nucleic acids.
- Spectroscopic and thermal properties of nucleic acids.

Likewise, it is advisable to know basic aspects of DNA replication, transcription and translation.

Regarding genes and gene expression, it is useful to have notions about:

- Genes.
- Genetic code.
- Central dogma of Molecular Biology.
- Gene expression



Learning outcomes

At the end of the course, the student must be able to prove that he/she has acquired the following learning outcomes:

- R1 Know the techniques used in Molecular Genetics.
- R2 Know the fundamental characteristics of the human genome and current genome sequencing methods.
- R3 Know the concepts around genetic variation.
- R4 Distinguish the different chromosomal abnormalities that may occur in humans.
- R5 Differentiate the different types of genetic alterations that cause disease.
- R6 Know the basics for the diagnosis of genetic diseases and is able to discern the right one for each case.



Competencies

Depending on the learning outcomes, the competencies to which the subject contributes are (please score from 1 to 4, being 4 the highest score):

BASIC		Weighting			
		1	2	3	4
CB1	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				X
CB2	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				X
CB3	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				X
CB4	Students can pass on information, ideas, problems and solutions to both a specialized and non-specialized audience				X
CB5	Students have developed the learning skills needed to undertake further studies with a high degree of autonomy				X
GENERAL		Weighting			
		1	2	3	4
CG1	Recognizing the essential elements of the medical profession, including ethical principles, legal responsibilities, and patient-centered professional exercise	X			
CG2	Understanding the importance of such principles for the benefit of the patient, society and profession, with special attention to professional secrecy		X		
CG3	Knowing how to apply the principle of social justice to professional practice and understanding the ethical implications of health in a changing global context	X			



CG4	Developing professional practice with respect to patient autonomy, beliefs and culture	X			
CG5	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		X		
CG6	Developing professional practice with respect for other health professionals, acquiring teamwork skills	X			
CG30	Basic knowledge of the National Health System and health legislation	X			
CG32	Knowing how to use information and communication technologies in clinical, therapeutic, preventive and research activities	X			
CG33	Maintaining and using records with patient information for further analysis, preserving data confidentiality	X			

SPECIFIC		Weighting			
		1	2	3	4
CE61	Assessing the risk-benefit ratio of diagnostic and therapeutic procedures			X	
CE62	Knowing the indications of biochemical, haematological, immunological, microbiological, anatomopathological and imaging tests				X
CE63	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		X		

TRANSVERSAL		Weighting			
		1	2	3	4
CT1	Analytical and synthesis capacity			X	
CT2	Planification and organization capacity		X		
CT6	Manage information capacity	X			



CT7	Solving problems				X		
CT8	Making decisions		X				
CT9	Team work				X		
CT10	Interdisciplinary team work		X				
CT12	Interpersonal relationship skills		X				
CT14	Critical reasoning					X	
CT16	Individual learning						X
CT18	Creativity		X				
CT19	Leadership		X				
CT24	Ability to take responsibility					X	
CT25	Autocriticism capacity					X	
CT26	Knowing how to value personal action and know your own skills and limitations					X	



Assessment system for the acquisition of competencies and grading system

Assessed learning outcomes	Granted percentage	Assessment method
R1, R3, R5	0,00%	Open questions
R1, R2, R3, R4, R5, R6	70,00%	Tests
R1, R3, R5, R6	30,00%	Practices
	0,00%	Participation in class

Observations

Remarks about the evaluation system

The evaluation instruments will be the following:

- Final exam (multiple-choice questions): 6 points
- Questionnaires (multiple-choice questions): 1 point (5 questionnaires, 0.2 points each). The questionnaires will be answered through the subject platform.
- Lab report: 1 point
- Practical exercises: 2 points

The top grade for the course will be 10 points. In order to pass the course, at least a 5-point grade must be achieved. The total grade will be calculated by adding the points obtained in each of the above mentioned evaluation instruments, as long as at least a 5-point grade (50%) is obtained in the final exam, which will contain questions about both theory and practice (lab and practice exercises) content .

Minimum requirements: You cannot take any subject of module 1 (Clinical Training Human) without having passed all the subjects of module 3 (Morphology, Structure and Function of the Human Body).

Criteria for awarding honors:

Honors will be granted to the best students achieving at least a 9-point grade. At most, only 1 student per 20 enrolled students (not per fraction of 20) may receive honors. If necessary, there will be special exam to determine those students deserving honors. In second and subsequent calls, honors may only be granted in case not all honors have been awarded in the first call.

Development of the subject in second and subsequent enrollments: There will be a specific group for students enrolled for second or more times if the total number of students exceeds the occupancy limit of the classroom, and a professor will be in charge of said group. The professor in



charge of this group will carry out 6 sessions of monitoring and tutoring of 2 hours each. The competencies to acquire the skills and of the subject will be carried out through all the practices planned for the course. In each session the subject will be developed in such a way to allow the students to acquire the competences needed to pass the subject. The evaluation of content and skills will be performed in the exam set in the official calendar for this course .

MENTION OF DISTINCTION:

According to Article 22 of the Regulations governing the Evaluation and Qualification of UCV Courses, the mention of "Distinction of Honor" may be awarded by the professor responsible for the course to students who have obtained, at least, the qualification of 9 over 10 ("Sobresaliente"). The number of "Distinction of Honor" mentions that may be awarded may not exceed five percent of the number of students included in the same official record, unless this number is lower than 20, in which case only one "Distinction of Honor" may be awarded.

Learning activities

The following methodologies will be used so that the students can achieve the learning outcomes of the subject:

M1	Masterclass
M2	Problems resolution and practical cases
M4	Content presentations by teacher
M5	Knowledges and skills explanation
M6	Laboratory practices
M7	Oral presentation by student
M8	Group activities supervised by professor
M9	Knowledge acquirance through student interaction and activity
M11	Personalised attention by professor
M12	Tests to understand the level of knowledge acquirance and skills



- M13 Written work
- M14 Online activity on e-learning
- M15 Personal study
- M16 Information research
- M17 Discussion and solving issues in group
- M18 Work in team
- M19 Group work for searching, discussion and information research
- M21 Supervision of clinical histories



IN-CLASS LEARNING ACTIVITIES

	LEARNING OUTCOMES	HOURS	ECTS
Theory class M1, M2, M4, M5, M9	R1, R2, R3, R4, R5, R6	29,50	1,18
Seminar and group practices M5, M6, M11	R1, R3, R5, R6	4,00	0,16
Tutoring M11	R1, R2, R3, R4, R5, R6	2,00	0,08
Evaluation M12	R1, R2, R3, R4, R5, R6	2,00	0,08
TOTAL		37,50	1,50

LEARNING ACTIVITIES OF AUTONOMOUS WORK

	LEARNING OUTCOMES	HOURS	ECTS
No attendance M2, M15	R1, R2, R3, R4, R5, R6	37,50	1,50
TOTAL		37,50	1,50



Description of the contents

Description of the necessary contents to acquire the learning outcomes.

Theoretical contents:

Content block

Contents

SECTION I: THE HUMAN GENOME

Unit 1: Genetics and genomics in Medicine.

- 1.1 The practice of genetics.
- 1.2 Categories of genetic diseases.

Unit 2: Introduction to the genome human.

- 2.1 Key ideas and questions.
- 2.2 The human genome and the chromosomal bases of inheritance.
- 2.3 Variation in the human genome.
- 2.4 Transmission of the genome.
- 2.5 Human gametogenesis and fertilization.
- 2.6 Medical importance of mitosis and meiosis.

Unit 3: Structure and function of human genes.

- 3.1 Key ideas and questions.
- 3.2 The central dogma: DNA -> RNA -> protein.
- 3.3 Gene expression.
- 3.4 Variation of gene expression and its importance in medicine.



SECTION II: ALTERATION OF THE HUMAN GENOME

Unit 4: Human genetic diversity: mutation and polymorphism.

- 4.1 Key ideas and questions.
- 4.2 Origin and frequency of different types of mutations.
- 4.3 Types of mutations and their consequences.
- 4.4 Variation in individual genomes.

Unit 5: Clinical cytogenetics and genomic analysis.

- 5.1 Key ideas and questions.
- 5.2 Chromosome analysis.
- 5.3 Genomic analysis.
- 5.4 Chromosomal abnormalities.

Unit 6: Chromosomal and genomic bases of the disease: disorders of the autosomes and sex chromosomes.

- 6.1 Key ideas and questions.
- 6.2 Mechanisms of anomalies.
- 6.3 Aneuploidy.
- 6.4 Uniparental disomy.
- 6.5 Microdeletion and duplication syndromes.
- 6.6 Disorders associated with genomic imprinting.
- 6.7 Sex chromosomes and their abnormalities.

SECTION III: PATTERNS OF INHERITANCE

Unit 7: Monogenic inheritance.

- 7.1 Ideas and key questions.
- 7.2 Family trees.
- 7.3 Autosomal Mendelian inheritance.
- 7.4 X-linked inheritance.
- 7.5 Mosaicism.
- 7.6 Dynamic mutations: expansion of unstable repeats.
- 7.7 Inheritance of disorders due to mutations in the mitochondrial genome.

Unit 8: Complex inheritance.

- 8.1 Key ideas and questions.
- 8.2 Qualitative and quantitative features.
- 8.3 Examples of common multifactorial diseases with genetic contribution.
- 8.4 Examples of multifactorial traits with known specific genetic and environmental factors.



BLOCK IV: GENETIC AND MOLECULAR BASES OF HUMAN GENETIC DISEASES

Unit 9: Identification of the genetic bases of human diseases.

- 9.1 Key ideas and questions.
- 9.2 Genetic basis for linkage analysis and association.
- 9.3 Mapping of human genes causing diseases.
- 9.4 From gene mapping to gene identification.
- 9.5 Searching for genes by sequencing the genome

Unit 10: Molecular bases of the genetic diseases.

- 10.1 Key ideas and questions.
- 10.2 How mutations affect synthesis and function of proteins.
- 10.3 Relationship between genotype and phenotype.

SECTION V: GENETICS AND GENOMICS OF CANCER

Unit 11: Genetics and genomics of cancer.

- 11.1 Key ideas and questions.
- 11.2 Genetic bases of cancer.
- 11.3 Familial cancer.
- 11.4 Sporadic cancer.
- 11.5 Cancer and the environment.

SECTION VI: CLINICAL GENETICS

Unit 12: Risk assessment and genetic counseling.

- 12.1 Family history in risk assessment.
- 12.2 Genetic counseling in clinical practice.
- 12.3 Empirical recurrence risks.
- 12.4 Molecular and genome-based diagnosis.

Unit 13: Prenatal diagnosis.

- 13.1 Prenatal diagnostic methods.
- 13.2 Laboratory tests.
- 13.3 Genetic counseling for prenatal diagnosis.

Unit 14: Ethical aspects in genetics and genomics.

- 14.1 Principles of biomedical ethics.
- 14.2 Ethical dilemmas in medical genetics.
- 14.3 Eugenic and dysgenetic effects of genetics medical.



Temporary organization of learning:

Block of content	Number of sessions	Hours
SECTION I: THE HUMAN GENOME	3,00	6,00
SECTION II: ALTERATION OF THE HUMAN GENOME	4,00	8,00
SECTION III: PATTERNS OF INHERITANCE	4,75	9,50
BLOCK IV: GENETIC AND MOLECULAR BASES OF HUMAN GENETIC DISEASES	4,00	8,00
SECTION V: GENETICS AND GENOMICS OF CANCER	2,00	4,00
SECTION VI: CLINICAL GENETICS	1,00	2,00

References

Basic bibliography

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.

Complementary bibliography
NOVO, F.J., Genética Humana: conceptos, mecanismos y aplicaciones de la genética en el campo de la Biomedicina, Pearson Prentice Hall, 2007.



Addendum to the Course Guide of the Subject

Due to the exceptional situation caused by the health crisis of the COVID-19 and taking into account the security measures related to the development of the educational activity in the Higher Education Institution teaching area, the following changes have been made in the guide of the subject to ensure that Students achieve their learning outcomes of the Subject.

Situation 1: Teaching without limited capacity (when the number of enrolled students is lower than the allowed capacity in classroom, according to the security measures taken).

In this case, no changes are made in the guide of the subject.

Situation 2: Teaching with limited capacity (when the number of enrolled students is higher than the allowed capacity in classroom, according to the security measures taken).

In this case, the following changes are made:

1. Educational Activities of Onsite Work:

All the foreseen activities to be developed in the classroom as indicated in this field of the guide of the subject will be made through a simultaneous teaching method combining onsite teaching in the classroom and synchronous online teaching. Students will be able to attend classes onsite or to attend them online through the telematic tools provided by the university (videoconferences). In any case, students who attend classes onsite and who attend them by videoconference will rotate periodically.

In the particular case of this subject, these videoconferences will be made through:

☒ Microsoft Teams

☐ Kaltura



Situation 3: Confinement due to a new State of Alarm.

In this case, the following changes are made:

1. Educational Activities of Onsite Work:

All the foreseen activities to be developed in the classroom as indicated in this field of the guide of the subject, as well as the group and personalized tutoring, will be done with the telematic tools provided by the University, through:

☒ Microsoft Teams

☐ Kaltura

Explanation about the practical sessions:



2. System for Assessing the Acquisition of the competences and Assessment System

ONSITE WORK

Regarding the Assessment Tools:

☒ The Assessment Tools will not be modified. If onsite assessment is not possible, it will be done online through the UCVnet Campus.

☐ The following changes will be made to adapt the subject's assessment to the online teaching.

Course guide		Adaptation	
Assessment tool	Allocated percentage	Description of the suggested changes	Platform to be used

The other Assessment Tools will not be modified with regards to what is indicated in the Course Guide.

Comments to the Assessment System: