

**COURSE DATA****Data Subject**

Code	36355
Name	Enfermedades raras
Cycle	Grade
ECTS Credits	4.5
Academic year	2024 - 2025

Study (s)

Degree	Center	Acad. Period	year
1204 - Degree in Medicine	Faculty of Medicine and Odontology	4	Second term

Subject-matter

Degree	Subject-matter	Character
1204 - Degree in Medicine	18 - Optional subjects	Optional

Coordination

Name	Department
ROMA MATEO, CARLOS	190 - Physiology
TORTAJADA GIRBES, MIGUEL	290 - Pediatrics, Obstetrics and Gynaecology

SUMMARY

The main goal in this optional subject 'Rare Diseases' is to create an academic and formative space, which provides students with tools for their future professional development, when facing an orphan disease. This subject intends to supply the lack of specific formation in this field, with special attention to the fact of addressing rare diseases in the learning process which students experience, as specific knowledge and skills are required. A formative strategy which comprises specific content regarding rare diseases is applied. It also considers measures the main of objective of which is to promote interest and sensitivity towards the problems these diseases cause.

PREVIOUS KNOWLEDGE



Relationship to other subjects of the same degree

There are no specified enrollment restrictions with other subjects of the curriculum.

Other requirements

Any degree studies would be enough to access epidemiological knowledge. However, their application in the field of rare diseases requires that students are familiar with aspects of biology and human physiopathology.

1204 - Degree in Medicine

- Students must be able to apply their knowledge to their work or vocation in a professional manner and have acquired the competences required for the preparation and defence of arguments and for problem solving in their field of study.
- Students must have the ability to gather and interpret relevant data (usually in their field of study) to make judgements that take relevant social, scientific or ethical issues into consideration.
- Understand and recognise the effects, mechanisms and manifestations of diseases over the structure and function of the human body.
- Recognise health determinants in population, such as genetic ones, dependent on sex, lifestyle, demographic, environmental, social, economic, psychological and cultural.
- Organizar y planificar adecuadamente la carga de trabajo y el tiempo en las actividades profesionales.
- Capacidad para trabajar en equipo y para relacionarse con otras personas del mismo o distinto ámbito profesional.
- Criticism and self-criticism skills.
- Capacity for communicating with professional circles from other domains.
- Acknowledge diversity and multiculturality.
- Consideration of ethics as a fundamental value in the professional practise.
- Working capacity to function in an international context.

1. Students will acquire knowledge about:

- a. The notion of rare disease.
- b. Different clinical aspects related to rare diseases.
- c. Localisation of resources and information about them.



- d. Aspects of prevention and early detection, and the techniques used for it: neonatal screening, genetic diagnosis, etc.
- e. Therapeutical aspects: advanced therapies, orphan drugs, contributing factors and healthcare products.
- f. Aspects regarding social issues and health care.
- g. New lines of research in the field.

2. Students will know the most relevant aspects of:

- a. General and specific characteristics of rare diseases.
- b. General and specific characteristics of orphan drugs.
- c. National and international legal framework, and policies applied within the field of rare diseases and orphan drugs.
- d. Main organisations, institutions, health portals and networks related to rare diseases.

3. Students will develop competencies of variable size in relation to the following:

- a. Associations of patients suffering from rare diseases.
- b. Coordination between levels of health care. Pharmaceutical care and pharmacotherapeutical follow-up in the field of rare diseases.

DESCRIPTION OF CONTENTS

1. THEORETICAL THEMES.

1. Rare Diseases: introduction and generalities. Epidemiology and general classification of rare diseases.
2. Bases of inheritance and Mendelian inheritance patterns. Knowledge of non-Mendelian models of inheritance (mitochondrial, sex-linked, genetic imprinting, etc.). Genetic polymorphisms and mutations. Concept of hereditary disease (penetrance, expressivity, genetic and phenotypic heterogeneity).
3. Molecular bases of rare diseases: diseases due to defects in the synthesis or catabolism of complex molecules; diseases due to the accumulation of toxic substances; energy deficiency diseases.
4. Strategy on Rare Diseases of the National Health System (health records, early detection and diagnosis). Legislation on rare diseases. Importance of biomedical research in rare diseases and information tools.
5. Congenital anomalies.
6. Rare infectious and parasitic diseases.
7. Rare nephrourological diseases.
8. Rare endocrine and metabolic diseases.
9. Rare diseases of the nervous system and sensory organs.



10. Rare diseases of the cardiovascular system.
11. Rare diseases of the respiratory system.
12. Rare diseases of the osteomyoarticular system and connective tissue.
13. Rare onco-hematological diseases.
14. Rare diseases of immune origin. Autoimmune diseases.
15. Rare diseases of the skin and subcutaneous tissue.
16. Orphan drugs: concept, specific problems, legislation and regulations.

2. PRACTICAL LESSONS

1. Presentation of clinical cases / works in the form of seminars, focused on specific topics on pathologies that will be taught during theory classes.
2. Attendance at workshops and talks on rare diseases, with the participation of specialist speakers in each field, with the following programming:

Workshop 1. Epidemiology and databases in Rare Diseases

Workshop 2. Patient associations

Workshop 3. Epigenetics and analysis of cases from the genetic point of view in Rare Diseases

Workshop 4. Biotechnology-based companies for the diagnosis of Rare Diseases

Workshop 5. Psychosocial aspects of Rare Diseases: role of primary care

WORKLOAD

ACTIVITY	Hours	% To be attended
Theory classes	19,00	100
Seminars	16,00	100
Computer classroom practice	10,00	100
Attendance at events and external activities	10,00	0
Development of group work	8,00	0
Development of individual work	10,00	0
Study and independent work	6,00	0
Readings supplementary material	4,00	0
Preparation of evaluation activities	6,00	0
Preparing lectures	8,00	0
Preparation of practical classes and problem	8,00	0
Resolution of case studies	4,00	0
Resolution of online questionnaires	3,50	0
TOTAL	112,50	



TEACHING METHODOLOGY

In the theoretical classes, the teacher will present, through a master lesson, the most important concepts and contents in a structured way, to obtain the knowledge and skills that the students must acquire. Student participation will be enhanced. The teaching material used by the teacher will be available, if the teacher considers it appropriate, from the electronic resource of the Virtual Classroom.

Classroom practices: seminars. In small groups the teacher will present in-depth specialized topics, case studies, bibliography management, current affairs... Group work and oral presentation will be encouraged. It could be understood as cooperative learning.

Clinical case study. Intensive and complete analysis of a real fact, problem or event in order to understand it, interpret it, contrast data, diagnose it and, sometimes, train in possible alternative solution procedures. Internships in Pediatric Dysmorphology Units, Genetic Services and associations of patients affected by rare diseases.

The gender perspective, the respect for diversity, and the sustainable development goals (SDGs) will be incorporated into teaching, whenever possible.

EVALUATION

The evaluation will be done continuously and all the training activities carried out (theory and practice) will be assessed.

The **THEORETICAL** part (50% of the final grade) includes the lessons taught during the lectures: they will be evaluated by means of a written test of 30 multiple-choice questions (without penalty for incorrect answer), with a total value of 5 points.

The **PRACTICAL** part (50% of the final grade) includes conducting seminars in team, which will be evaluated by oral presentation and defense of the works, and evaluated with a total of 4 points; and attendance at workshops / talks, which will be evaluated with 1 point.

The final grade will be the sum of both parts, theoretical and practical. Both must be approved separately, in such a way that a minimum of 2.5 points must be obtained in each part (theory and practical), being therefore necessary to take the theoretical exam to pass. The maximum grade for the subject will be 10 and the grade for the pass will be 5.

It is a requirement to access the advanced call for this subject that the student has completed all of their practices.

Attendance at the exhibition of practical work will be mandatory.

Attendance at practical activities is mandatory. The student is considered to meet this requirement if he or she has attended a minimum of 80% of these activities and has adequately justified the impossibility of attending the remaining sessions due to the occurrence of a cause of force majeure. It will be essential to comply with this requirement to pass the subject.



Students are reminded of the importance of carrying out evaluation surveys on all the teaching staff of the degree subjects.

REFERENCES

Basic

- IZQUIERDO M, AVELLANEDA A. Enfermedades Raras: Un enfoque práctico. 1a Edición. Madrid: ISCIII; 2004.
- Servicio de Pediatría. Hospital Universitario «Marqués de Valdecilla». Universidad de Cantabria. Necesidades de los pacientes pediátricos con enfermedades raras y de sus familias en Cantabria. Documentos 69/2005. Real Patronato sobre Discapacidad. Ministerio de Trabajo y Asuntos Sociales.
- POSADA DE LA PAZ M, GROFT STEPHEN C, eds. Rare diseases epidemiology. Series: Advances in Experimental Medicine and Biology, Vol. 686. 1ªed. Springer; 2010.
- Instituto de Investigación en Enfermedades Raras. ISCIII. Registro de Enfermedades Raras. Disponible en: <https://registorraras.isciii.es>
- KLEIN DE ZIGHELBOIM E, GALLARDO JUGO BE, CHÁVEZ PASTOR M, ABARCA BARRIGA HH. Atlas de Dismorfología Pediátrica. Fondo Editorial del Instituto Nacional de Salud del Niño. Lima, Perú 2012.
- Estrategia en enfermedades raras del Sistema nacional de Salud. Sanidad 2009. Ministerio de Sanidad y Política Social. Disponible en:

<https://www.mscbs.gob.es/organizacion/sns/planCalidadSNS/docs/enfermedadesRaras.pdf>
- Enfermedades raras y medicamentos huérfanos. Jules J. Berman. Elsevier 2015
- RECURSOS e-Salut:
 - ClinicalKey Student Medicina, Odontología y Enfermería
[<https://uv-es.libguides.com/RecursosSalut>]
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