

**COURSE DATA****DATA SUBJECT**

**Code:** 36355  
**Name:** Rare Diseases  
**Cycle:** Undergraduate Studies  
**ECTS Credits:** 4.5  
**Academic year:** 2025-26

**STUDY (S)**

Degree	Center	Acad. year	Period
1204 - Degree in Medicine	Facultat de Medicina i Odontologia	4	Second quarter

**SUBJECT-MATTER**

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

**COORDINATION**

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**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 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.

## COMPETENCES / LEARNING OUTCOMES

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Acknowledge diversity and multiculturality.

Capacity for communicating with professional circles from other domains.

Consideration of ethics as a fundamental value in the professional practise.

Criticism and self-criticism skills.

Proper organisation and planning of the workload and timing in professional activities.

Recognise health determinants in population, such as genetic ones, dependent on sex, lifestyle, demographic, environmental, social, economic, psychological and cultural.

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.

Team-working skills and engaging with other people in the same line of work or different.

Understand and recognise the effects, mechanisms and manifestations of diseases over the structure and function of the human body.

Working capacity to function in an international context.

## DESCRIPTION OF CONTENTS

### 1. THEORETICAL THEMES.

1. Rare Diseases: Introduction and generalities. Epidemiology and general classification of rare diseases.

2. Basis of inheritance and Mendelian inheritance patterns. Knowledge of non-Mendelian inheritance models. Genetic polymorphisms and mutations.



3. Molecular basis of rare diseases: diseases due to defects in the synthesis or catabolism of complex molecules; diseases due to the accumulation of toxic substances; diseases due to energy deficiency.
4. Rare Disease Strategy of the National Health System (health registries, early detection and diagnosis). Legislation on rare diseases. Importance of biomedical research in rare diseases and information tools.
5. Orphan drugs: concept, specific problems, legislation, and regulations.
6. Congenital anomalies.
7. Rare infectious and parasitic diseases.
8. Rare nephro-urological diseases.
9. Rare endocrine and metabolic diseases.
10. Rare diseases of the nervous system and sensory organs.
11. Rare diseases of the cardiovascular system.
12. Rare diseases of the respiratory system.
13. Rare diseases of the osteo-myo-articular system and connective tissue.
14. Rare onco-hematological diseases.
15. Rare diseases of immune origin. Autoimmune diseases.
16. Rare diseases of the skin and subcutaneous tissue.

## **2. PRACTICAL LESSONS**

1. Presentation of clinical cases/assignments in the form of seminars, focusing on specific topics related to the pathologies that will be taught during the theory classes. Work in groups, grading will be assessed through submission of work and in-person presentation.

2. Attendance at workshops and talks on rare diseases, with the participation of specialist speakers in each field, with the following program:

Workshop 1. Epidemiology and Databases in Rare Diseases



Workshop 2. Patient Associations

Workshop 3. Case Analysis from a Genetic Perspective in Rare Diseases

Workshop 4. Biotechnology-Based Companies for the Diagnosis of Rare Diseases

Workshop 5. Psychosocial Aspects of Rare Diseases: The Role of Primary Care

3. Rare Disease Book Club - group discussion activity divided into two sessions:

Session 1. Presentation of the comic book(s) belonging to the Graphic Medicine movement; preliminary survey.

Session 2. Debate and exchange of opinions after the reading; final survey.

## WORKLOAD

### PRESENCIAL ACTIVITIES

Activity	Hours
Theory	19,00
Seminar	16,00
Computer classroom practice	10,00
<b>Total hours</b>	<b>45,00</b>

### NON PRESENCIAL ACTIVITIES

Activity	Hours
Attendance at other activities	10,00
Individual or group project	18,00
Independent study and work	10,00
Preparation of lessons	16,00
Preparation for assessment activities	9,50
Resolution of case studies	4,00
<b>Total hours</b>	<b>67,50</b>

## 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 Dismorphology 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

Assessment will be continuous and all training activities completed (theory and practical) will be evaluated.

The theoretical part (50% of the final grade) includes the lessons taught during class: they will be assessed by a written test of 30 multiple-choice questions (with a penalty for incorrect answers), worth a total of 5 points.

The practical part (50% of the final grade) includes team seminars, which will be assessed by oral presentations and presentation of the work, earning a total of 4 points; and the completion of a final report/test after the debate of the reading club, which will be worth 1 point. This test will be administered in the same session as the theoretical content exam.

The final grade will be the sum of both the theoretical and practical sections. Both must be passed separately, with a minimum score of 2.5 points required in each section (theory and practical). Therefore, students must take the theoretical exam to pass. The maximum grade for the subject is 10, and the passing grade is 5. Students must complete all practical activities to advance the call for this subject.

Attendance at the practical presentation is mandatory. Attendance at practical activities is mandatory. Students are considered to meet this requirement if they have attended at least 80% of these activities and have adequately justified their inability to attend the remaining sessions due to a force majeure event. This requirement must be met to pass the subject.

Students are reminded of the importance of completing the evaluation surveys for all faculty members of the degree courses.

## REFERENCES



- 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.
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- Estrategia en enfermedades raras del Sistema nacional de Salud. Sanidad 2009. Ministerio de Sanidad y Política Social. Disponible en: <https://www.msbs.gob.es/organizacion/sns/planCalidadSNS/docs/enfermedadesRaras.pdf>
- Enfermedades raras y medicamentos huérfanos. Jules J. Berman. Elsevier 2015
- Pediatría y enfermedades raras. Enfermedades lisosomales. Casos clínicos Coordinador: Luis González Gutiérrez-Solana. Neuropediatría, Unidad de Neurodegenerativas, Hospital Infantil Universitario Niño Jesús, Madrid. Ergón Creación, S.A. (ISBN: 978-84-16732-40-1)
- Ética en la investigación de las Enfermedades Raras; Carmen Ayuso García, Rafael Dal-Ré Saavedra, Francesc Palau Martínez. Ergón Creación, S.A. (ISBN: 978-8416270859)
- Enfermedades de impronta: Guías de buena práctica clínica. Editado por Guiomar Pérez de Nanclares y Pablo Lapunzina (ISBN: 978-8416270859)
- Epiléptico. El ascenso del Gran Mal. David B. Salamandra Graphic (ISBN: 978-8416131488).
- Ausencias. Ramón Rodríguez y Cristina Bueno. Astiberri (ISBN: 978-84-15163-76-3).
- RECURSOS e-Salut:
  - ClinicalKey Student Medicina, Odontología y Enfermería [<https://uv-es.libguides.com/RecursosSalut>]
  - Acces Medicina [[https://uv-es.libguides.com/Access\\_Medicina](https://uv-es.libguides.com/Access_Medicina)]
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