# COURSE DATA

| Data Subject | |
|---|---|
| **Code** | 36355 |
| **Name** | Enfermedades raras |
| **Cycle** | Grade |
| **ECTS Credits** | 4.5 |
| **Academic year** | 2022 - 2023 |

**Study (s)**

| Degree | Center | Acad. year | Period |
|---|---|---|---|
| 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.

## OUTCOMES

### 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.
- Proper organisation and planning of the workload and timing in professional activities.
- Team-working skills and engaging with other people in the same line of work or different.
- 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.

## LEARNING OUTCOMES

**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 general aspects. Epidemiology. General classification of rare diseases.
2.Bases of heredity and patterns of Mendelian Inheritance. Knowledge of the non-Mendelian models of Inheritance (mitochondrial, sex-linked, genomic imprinting, etc.). Genetic polymorphisms and mutations. The concept of inherited disease (penetrance, expressivity, genetic and phenotypic heterogeneity).
3.Molecular bases of rare diseases. Diseases due to a defect in the synthesis or catabolism of complex molecules: lysosomal and peroxisomal diseases, diseases of the intracellular transportation and processing. Diseases due to accumulation of toxic substances: aminoacidopathies, organic aciduria, disorders of the urea cycle, sugar intolerance. Diseases due to energy-deficit, such as glycogen storage disease, defects in the glugoneogenesis, congenital lactic academia, oxidation disorders of fatty acids, and diseases of the mitochondrial respiratory chain.
4.Strategy for Rare Diseases in the National Health Care System (health records, early detection and diagnosis). Rare diseases legislation. The importance of biomedical research on rare diseases.

Information tools on rare diseases (Orphanet, Eurodis).
5.Congenital anomalies.
6.Parasitic and infectious rare diseases.
7.Nephro-urological rare diseases.
8.Endocrine and metabolic rare diseases.
9.Rare diseases of the nervous system and sense organs.
10.Rare diseases of the cardiovascular system.
11.Rare diseases of the respiratory system.
12.Rare diseases of the osteo-myoarticular system and the connective tissue.
13.Rare onco-haematological diseases.
14.Rare diseases of immunitary origin. Autoimmune diseases.
15.Rare diseases of skin and subcutaneous tissue.
16.Psychosocial aspects of RD.
17.Therapeutical aspects of RD. The concept of orphan drugs.

### 2. PRACTICAL LESSONS

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

Workshop 1. Epidemiology and databases in Rare Diseases
Workshop 2. Patient associations
Workshop 3. Epigenetics of RD
Workshop 4. Biotechnology-based companies for the diagnosis of RD
Workshop 5. Psychosocial aspects of RD

## 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

Regarding theoretical credits, professors will present the most important methods, procedures and content through master classes, in order to help students develop several skills and meet the objectives. Students' participation in the class should be encouraged and they will have access to the didactic material professors may have used through the electronic platform *Aula Virtual*.

Practise in the classroom: seminaries. In small groups, professors will present deeply specialised themes, case studies, management of bibliography, current issues… Team work is encouraged, as well as oral presentation skills, which could be interpreted as 'cooperative learning'.

Clinical case studies. Intensive and complete analysis of a fact, a problem or a real situation in order to know about it, how to interpret it, contrast data, make a diagnosis and, in some occasions, be trained on how to apply possible alternative solutions.

Practise in Units of Paediatric Dysmorphology, Medical Genetic Services and Associations of patients suffering from rare diseases.

## 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 to practical sessions is mandatory. Unjustified non-attendance to more than 20% of the sessions will make it impossible to pass the course.

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 Valdecila». 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.
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- 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. Elsevier (Scopus, ScienceDirect): uv-es.libguides.com/RecursosSalut/BibliotecaSalut