COURSE DATA

DATA SUBJECT

Code: 43085
Name: Physiopathology of rare diseases
Cycle: Master's Degree
ECTS Credits: 4
Academic year: 2026-27

STUDY (S)

Degree	Center	Acad. year	Period
2141 - Master's Degree in Physiology	Facultat de Medicina i Odontologia	1	Second quarter

SUBJECT-MATTER

Degree	Subject-matter	Character
2141 - Master's Degree in Physiology	Oxidative stress and its applications in biomedicine	COMPULSORY

COORDINATION

PALLARDO CALATAYUD FEDERICO VICENTE

SUMMARY

In this subject, the pathophysiology of some rare diseases associated with oxidative stress is studied. Basic aspects about rare diseases, the problems inherent to their study, diagnosis and treatment, with special emphasis on the social aspects derived from them, will be initially taught. Next, examples of several rare diseases will be studied. Most of the professors will be medical doctors and guest researchers who are specialists in the field.

PREVIOUS KNOWLEDGE

RELATIONSHIP TO OTHER SUBJECTS OF THE SAME DEGREE

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

OTHER REQUIREMENTS

It is recommended that students take subjects in Physiology, Biochemistry and Molecular Biology and in topics related to Pathology, such as Pathophysiology and General Pathology.

COMPETENCES / LEARNING OUTCOMES

2141 - Master's Degree in Physiology

Assess the need to complete the scientific training, in languages, computer science, ethics, etc., attending conferences or courses and/or carrying out complementary activities, self-evaluating the contribution that the performance of these activities implies for their comprehensive training.

Know how to write and prepare presentations to present and defend them later.

Obtain new skills for the diagnosis and treatment of rare diseases, as well as their limitations, especially in those diseases that are genetically unstable and predisposed to cancer.

Search, order, analyze and synthesize scientific information (databases, scientific articles, bibliographic repertoires), selecting the pertinent to focus current knowledge on a topic of scientific interest in Physiology.

Students should apply acquired knowledge to solve problems in unfamiliar contexts within their field of study, including multidisciplinary scenarios.

Students should be able to integrate knowledge and address the complexity of making informed judgments based on incomplete or limited information, including reflections on the social and ethical responsibilities associated with the application of their knowledge and judgments.

Students should communicate conclusions and underlying knowledge clearly and unambiguously to both specialized and non-specialized audiences.

Students should demonstrate self-directed learning skills for continued academic growth.

Students should possess and understand foundational knowledge that enables original thinking and research in the field.

DESCRIPTION OF CONTENTS

1. Introduction to the study of rare diseases

Introduction and general aspects of rare diseases. Epidemiology of Rare Diseases.

2. Molecular bases of rare diseases

The role of mitophagy and mitochondrial dynamics in mitochondrial neuropathy. Role of mitochondrial DNA mutations.

3. Lafora disease, Dravet Disease and other epilepsies.

Malaltia de Lafora, Malaltia de Dravet y altres epilepsies. Característiques. Clinical profile. Physiopathology. Potential treatments.

4. Polyglutamine related diseases.

Huntington disease. Cerebellar ataxias. Clinical pattern. Etiology. Physiopathology. Therapeutical approaches.

5. Friedreich Ataxia.

Handling of a patient in a clinical ataxia unit. Diagnostic algorithm. Physiopathology. New treatments and future perspectives.

6. Down Syndrome

The most common rare disease. Clinical pattern. Physiopathology. Oxidative stress. New treatments for the future.

7. Pulmonary fibrosis

Epidemiology. Clinical pattern. Etiology. Physiopathology of the disease. Current therapy and future treatments.

8. Progerias

Types. Description of the disease. Etiology Physiopathology. Treatment. Role of oxidative stress.

9. Genodermatosis

Classification. Etiology. Physiopathology. Some examples. Treatments

10. Epigenetic bases of some rare diseases. Definition and main regulatory epigenetic mechanisms

General an particular aspects. Pathology of epigenetics. Diseases of epigenetic origin. Some examples. Current studies.

11. New therapeutics for the treatment of rare diseases

Orphan drugs. Clinical trials with rare diseases.

WORKLOAD

PRESENCIAL ACTIVITIES

Activity	Hours
Tutorials	3,00
Theory	24,00
Other activities	0,00
Total hours	**27,00**

NON PRESENCIAL ACTIVITIES

Activity	Hours
Attendance at other activities	2,00
Individual or group project	20,00
Independent study and work	20,00
Preparation of lessons	6,00
Preparation for assessment activities	15,00
Resolution of case studies	10,00
Total hours	**73,00**

TEACHING METHODOLOGY

- Theoretical classes.
- Conferences by experts in the field.
- Debate and guided discussion on the work carried out.
- Face-to-face and electronic tutorials with teachers.

EVALUATION

Evaluation system:

- Written exam. Multiple choice questions: evaluation up to 10 points.

Minimum passing grade: 5 points.

REFERENCES

- - http://www.orpha.net/consor/cgi-bin/index.php?lng=ES - http://www.ciberer.es -Orphanet Activity Report 2018 (disponible en https://www.orpha.net/orphacom/cahiers/docs/GB/ActivityReport2018.pdf - http://www.ciberer.es - Enfermedades raras y medicamentos huérfanos de Jules J. Berman, Elsevier España, S.L.U. (9788490229194) ISBN: 8490229198 ISBN-13: 9788490229194 - Epigenetic Biomarkers and Diagnostics (English Edition) . Dr. José Luis García Giménez (Editor). Academic Press. ISBN de origen : 0128018992

- Cada profesor aportará para su tema referencias complementarias.