

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

Code	43085
Name	Physiopathology of rare diseases
Cycle	Master's degree
ECTS Credits	4.0
Academic year	2021 - 2022

Study (s)

Degree	Center	Acad. year	Period
2141 - M.U. en Fisiología 12-V.2	Faculty of Medicine and Odontology	1	Second term

Subject-matter

Degree	Subject-matter	Character
2141 - M.U. en Fisiología 12-V.2	3 - Oxidative stress and its applications in biomedicine	Obligatory

Coordination

Name	Department
DASI FERNANDEZ, FRANCISCO JOSE	190 - Physiology

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.

OUTCOMES

2141 - M.U. en Fisiología 12-V.2

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

LEARNING OUTCOMES

To know the pathophysiology of the examples of rare diseases studied.

To learn about the role of free radicals, oxidative stress and redox signaling in the pathophysiology of rare diseases.

To learn about new therapeutic approaches for the rare diseases studied.

To learn about the social aspects of rare diseases.



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

Basic genetics. Mendelian and non-Mendelian heritage. Hereditary diseases. Introduction to oxidative stress.

3. Rare diseases of the nervous system

Pathophysiology. Diagnosis. Treatment. Oxidative stress and redox signalling. New lines of research.

4. Neurodegenerative disorders affecting motor function: ALS and MS in children

Pathophysiology. Diagnosis. Treatment. Oxidative stress and redox signaling. New lines of research.

5. Rare diseases of the respiratory system

Pathophysiology. Diagnosis. Treatment. Oxidative stress and redox signaling. New lines of research.

6. Rare diseases of gastrointestinal system

Pathophysiology. Diagnosis. Treatment. Oxidative stress and redox signaling. New lines of research.

7. Rare diseases of the osteo-articular system

Pathophysiology. Diagnosis. Treatment. Oxidative stress and redox signaling. New lines of research.

8. Rare diseases of the skin

Pathophysiology. Diagnosis. Treatment. Oxidative stress and redox signaling. New lines of research.

9. Rare diseases of the renal system

Pathophysiology. Diagnosis. Treatment. Oxidative stress and redox signaling. New lines of research.

**10. New therapeutic strategies for the treatment of rare diseases**

Orphan drugs. Clinical trials with Rare Diseases.

11. Communication and rare diseases

Importance of media in the field of rare diseases.

12. Psychosocial aspects of rare diseases

Psychosocial aspects. Patients' associations.

WORKLOAD

ACTIVITY	Hours	% To be attended
Theory classes	24,00	100
Tutorials	3,00	100
Other activities	2,00	100
Development of individual work	20,00	0
Study and independent work	15,00	0
Readings supplementary material	5,00	0
Preparation of evaluation activities	15,00	0
Preparing lectures	6,00	0
Resolution of case studies	10,00	0
TOTAL	100,00	

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

Basic

- <https://www.orpha.net/consor/cgi-bin/index.php>

Additional

- Cada profesor aportará para su tema referencias complementarias.

ADDENDUM COVID-19

This addendum will only be activated if the health situation requires so and with the prior agreement of the Governing Council

ONLY IF FACE-TO-FACE TEACHING AND EVALUATION IS NOT POSSIBLE:

1. Contents

The contents collected in the teaching guide are maintained.

2. Workload and temporary planning of teaching

The weight of the various activities that add up the hours of dedication in ECTS credits marked in the teaching guide is maintained.

Scheduled teaching dates and times are maintained.

3. Teaching methodology

Both theoretical topics and tutorials will take place virtually.

4. Evaluation

The evaluation system of the teaching guide is maintained. The exam will be made online on the day and time provided for in the exam schedule approved in the degree.

5. Bibliography

The bibliography recommended in the teaching guide is maintained.