Universidad Zaragoza

Academic Year/course: 2023/24

26739 - Rare Diseases in Children

Syllabus Information

Academic year: 2023/24
Subject: 26739 - Rare Diseases in Children
Faculty / School: 104 - Facultad de Medicina
Degree: 304 - Degree in Medicine
ECTS: 4.0
Year: 5
Semester: First semester
Subject type: Optional
Module:

1. General information

Rare diseases generally manifest themselves in the paediatric age (>80% of cases), and most of them are of genetic origin (90%). It is therefore appropriate that this subject is taught in the same term as the subject Paediatrics.

The general objectives of the subject are:

1. To know the existence and importance of rare diseases as a health and social-health problem.
2. To know the concepts and general aspects of rare diseases as a whole.
3. To know the clinical, diagnostic, therapeutic aspects, as well as the natural history of the main rare diseases in children.
4. To learn about real clinical cases of patients with rare diseases.

These approaches and objectives are aligned with the "Sustainable Development Goals" (SDGs) of the United Nations Agenda 2030 (https://un.org/sustainabledevelopment/es/). Acquisition of the subject learning results provides training and competence to contribute to their achievement:

Goal 3: Health and wellness

Goal 10: Reduction of inequalities

2. Learning results

The student, in order to pass this subject, must demonstrate the following results...

1. To know the concept and definition of rare disease.
2. To know the global prevalence of rare diseases in our environment (Spain, Europe).
3. To know the clinical/therapeutic generalities of rare diseases.
4. To know the main aspects of the Plans and Strategies (national and European) on rare diseases.
5. To know the importance of rare diseases in Paediatrics.
6. To know the main resources available for the study, research and social and health care of patients and families with rare diseases.
7. To know the clinic, diagnosis and treatments of some of the main rare diseases in the paediatric age.

Importance of learning results

Having taken this subject, the student (future health professional) will be aware of the existence and importance of rare diseases as a priority health problem in our health system and of the importance of the new genomic tools for diagnosis, multidisciplinary management and treatment.

3. Syllabus

THEORETICAL PROGRAM

- Introduction and General
- Turner syndrome
- Microdeletion syndromes
- X Fragile Syndrome
- Noonan syndrome. Rasopathies
- Inborn Errors of Metabolism
- Spinal Muscular Atrophy type 1

- Prader-Willi syndrome
- Cornelia de Lange syndrome
- Rett syndrome
- Myotonic Dystrophy type 1
- Mitochondrial diseases

CLINICAL CASE STUDIES:

Each of the diseases or group of diseases included in the theoretical program will be followed by the presentation and discussion of a real clinical case. Some of them will be attended by one or both parents of an affected patient.

4. Academic activities

The learning process that has been designed for this subject is based on the following :

- Face-to-face activities (25 hours)

- Non-face-to-face activities (75 hours)

1. Master classes (theoretical): 1 of 2 hours and 11 of 1 hour (12 classes = 13 hours).

2. Practical seminars (real practical clinical cases) of 1 hour; (11 seminars = 11 hours).

3. Completion of a written test, with 5 options and only 1 correct answer, without penalising wrong answers (1 hour) 4. Uni-personal work on any of the rare diseases included, or not, in the Program of the Subject, or on any general aspect related to them (75 hours).

5. Assessment system

The student must demonstrate achievement of the intended learning results through the following assessment activities:

1. Written exam (multiple choice): **60%** of the final grade: 20 questions with 5 options and only one correct answer, no penalisation for wrong answers. It is passed with 60% success rate

2. Practical seminars (clinical cases): **20%** of the final grade

3. Supervised work: **20%** of the final grade