

## 26739 - Rare Diseases in Children

### Syllabus Information

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**Academic Year:** 2019/20

**Subject:** 26739 - Rare Diseases in Children

**Faculty / School:** 104 -

**Degree:** 304 - Degree in Medicine

**ECTS:** 4.0

**Year:** 5

**Semester:** First semester

**Subject Type:** Optional

**Module:** ---

## 1.General information

### 1.1.Aims of the course

The subject and its expected results correspond to the following approaches and objectives:

Approach:

Rare diseases usually manifest themselves in the pediatric age (> 80% cases), being also the majority of genetic origin (?90%). It is therefore opportune that this subject be taught in the same course as the subject Pediatrics.

The general objectives of the subject are:

1. To know the existence and importance of rare diseases as a health and socio-sanitary 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 childhood diseases.
4. Know actual clinical cases of patients with rare diseases.

### 1.2.Context and importance of this course in the degree

As previously stated, rare diseases appear mostly in the pediatric age, in some cases already from birth (congenital). Its early diagnosis is essential for an appropriate treatment or therapeutic management.

The student of Medicine and other Degrees in Health Sciences must know the existence of rare diseases, recognizing the main symptoms, forms of diagnosis, treatment and natural history of the most prevalent. They must also be aware of the importance of these diseases as a priority problem in our National Health System.

### 1.3.Recommendations to take this course

It is advisable, although not essential, to be enrolled in any of the following Degrees:

- Medicine
- Nursing
- Physiotherapy
- Occupational Therapy

## 2.Learning goals

### 2.1.Competences

Upon passing the subject, the student will be competent to ...

1. Recognize the importance of rare diseases as a priority public health problem.
2. Know the general and common aspects of rare diseases.
3. To know the clinical, diagnostic, therapeutic and natural aspects of some of the most prevalent rare diseases in childhood.

### 2.2.Learning goals

The student, to overcome this subject, must demonstrate the following results:

- Know the concept and definition of rare disease.
- Know the global prevalence of rare diseases in our environment (Spain, Europe).
- Know the clinical / therapeutic generalities of rare diseases.
- Know the main aspects of the Plans and Strategies (National and European) on rare diseases.
- Know the importance of rare diseases in Pediatrics.
- Know the main resources available for study, research and health care for patients and families with rare diseases.
- Know the symptoms, diagnosis and treatments of the main rare diseases in the pediatric age.

### 2.3.Importance of learning goals

Having completed this course, the future health professional will be aware of the existence and importance of rare diseases as a priority health problem in our health system

## 3.Assessment (1st and 2nd call)

### 3.1.Assessment tasks (description of tasks, marking system and assessment criteria)

The student must demonstrate that he / she has attained the expected learning outcomes through the following evaluation activities:

1. Written exam (multiple choice questions, MCQ): 60% of the final mark
2. Practical seminars (clinical cases): 20% of the final mark
3. Guided work: 20% of the final grade

Scheduled dates for global assessments:

<https://medicina.unizar.es/quinto-curso#horario9>

## 4.Methodology, learning tasks, syllabus and resources

### 4.1.Methodological overview

The methodology followed in this course is oriented towards the achievement of the learning objectives. A wide range of teaching and learning tasks are implemented, such as lectures:

1. Lectures (theoretical) 1 hour (12 classes = 13 hours; 1st day 2 hours)
2. Practical Seminars (actual clinical cases) 1 hour; (11 classes = 11 hours)
3. Performing a written test (multiple choice, with 5 options and only one correct answer, no negatives for incorrect answers)
4. Autonomous work on some of the rare diseases included in the Syllabus, or any other (general) aspects of Rare Diseases.

### 4.2.Learning tasks

The course includes the following learning tasks:

- Attendance to lectures
- Attendance and active discussion of clinical cases
- Performing autonomous work related to rare diseases

### 4.3.Syllabus

The course will address the following topics:

- Topic 1. Introduction to Rare Diseases
- Topic 2. Turner Syndrome
- Topic 3. Microdeletion Syndromes
- Topic 4. Fragile X Syndrome
- Topic 5. Noonan syndrome: Rasopathies
- Topic 6. Menkes syndrome
- Topic 7. Inborn Errors of Metabolism
- Topic 8. Spinal Muscular Atrophy type 1
- Topic 9. Prader-Willi Syndrome
- Topic 10. Cornelia de Lange Syndrome
- Topic 11. Myotonic Dystrophy type 1
- Topic 12. Rett syndrome

### 4.4.Course planning and calendar

Scheduled lectures and presentation of works:

The lectures (theoretical lesson + clinical case) will be taught from 17:00 to 19:00 on the Monday lectures of the 9th semester

of the Academic Year. In case of being non-school, an alternative week-day will be searched.

The teaching activities of the subject will be taught in the time zone from 5:00 pm to 7:00 pm. On the Monday of the first semester of 5th year of the Degree (9th semester).

The teaching activity will be divided into:

- Face-to-face teaching (37.5%): 1.5 ECTS (37.5 hours) and
- non-face-to-face teaching (62.5%): 2.5 ECTS (62.5 hours).

The classroom teaching is divided between theoretical classes (13 hours) and practical clinical cases (11 hours). The key dates of the course will be set within the academic calendar established by the Center for each academic year. The examinations for the academic year will be the ones corresponding to January and September.

Scheduled (tentative) dates for evaluations:

<https://medicina.unizar.es/quinto-curso#horario9>

- First call: Decembre /January

- Second call: September

#### **4.5.Bibliography and recommended resources**

<http://psfunizar7.unizar.es/br13/eBuscar.php?tipo=a>