

Academic Year/course: 2022/23

26739 - Rare Diseases in Children

Syllabus Information

Academic Year: 2022/23

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

1.1. Aims of the course

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

A p p r o a c h :

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.

These approaches and objectives are aligned with the following "Sustainable Development Goals" (SDG) of the United Nations 2030 Agenda (<https://un.org/sustainabledevelopment/es/>), in such a way that the acquisition of learning outcomes of the subject provides training and competence to contribute to some extent to your **a c h i e v e m e n t :**

- Objective 3: Health and well-being
- Objective 10: Reduction of inequalities

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

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 mark

Scheduled dates for global assessments:

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

4. Methodology, learning tasks, syllabus and resources

4.1. Methodological overview

1. Master classes (theoretical): 1 of 2 hours and 11 of 1 hour (12 classes = 13 hours).
2. Practical seminars (practical real clinical cases) of 1 hour; (11 seminars = 11 hours).
3. Carrying out a written exam (1 hour) (MCQ type, with 5 options and only 1 correct answer, without negatives).
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

Face-to-face activities (25 hours = 2.5 ECTS):

- Attendance to theoretical lessons (13 hours = 1.3 ECTS)
- Attendance and participation in the discussion of clinical cases (11 hours = 1.1 ECTS)
- Taking the theoretical exam (1 hour = 0.1 ECTS)

Non-contact activities (75 hours = 7.5 ECTS):

- Carrying out (uni)personal work related to rare diseases (topic selection, design, development, bibliography and presentation)

NOTE: If for reasons of public health risk, the face-to-face teachings could not be given as planned, The guidelines of the Order of July 6, 2020 of the Governing Council of the UZ, by which they are adopted, would be followed guidelines to face the development of teaching in the academic year 2020-21, in the situation of new normality.

4.3. Syllabus

The course will address the following topics*:

- Topic 1. Introduction and general overview on Rare Diseases
- Topic 2. Turner Syndrome
- Topic 3. Microdeletion Syndromes
- Topic 4. Fragile X Syndrome
- Topic 5. Noonan syndrome. Rasopathies
- Topic 6. Inborn Errors of Metabolism

- Topic 7. Spinal Muscular Atrophy type 1
- Topic 8. Prader-Willi Syndrome
- Topic 9. Cornelia de Lange Syndrome
- Topic 10. Rett Syndrome
- Topic 11. Myotonic Dystrophy type 1
- Topic 12. Mitochondrial diseases

* The order of delivery may vary depending on the availability of professors.

4.4. Course planning and calendar

Calendar of face-to-face sessions and presentation of work

The face-to-face classes (theoretical lesson + clinical case) will be taught from 5:00 p.m. to 7:00 p.m. on Monday of the 9th semester of the Academic Course. In case of being non-teaching or due to manifest impossibility, an alternative working day will be sought.

The face-to-face teaching activities of the subject will be taught from 5:00 p.m. to 7:00 p.m. on Monday classes of the first semester of the 5th year of the Degree (9th semester). The key dates of the course and of the exam will be set within the academic calendar established by the Center for each academic year.

Scheduled (tentative) dates for evaluations:

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

4.5. Bibliography and recommended resources

<http://psfunizar10.unizar.es/br13/egAsignaturas.php?codigo=26739>