

26739 - Unusual Children's Illnesses

Información del Plan Docente

Academic Year	2017/18
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.Introduction

Rare diseases are currently an important group of pathologies that, due to their infrequent condition, require a multidisciplinary approach at both the medical and socio-sanitary levels.

Given their low frequency, it is common for both medical professionals and professionals in the socio-sanitary area to lack the experience necessary for their correct approach, management and, if available, treatment.

It is for this reason that the corresponding health authorities, both at the regional, national and European levels, have tackled the problem in a global way to advance their knowledge and therapeutics, as well as to establish specific resources to investigate in all aspects related to them (medical, social, etc ...)

All of the aforementioned, together with the need for basic medical care to affected patients, makes the inclusion of this topic in the medical curricula of study justified.

1.2.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

1.3.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.

1.4.Activities and key dates

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

26739 - Unusual Children's Illnesses

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 / February and September.

The initial dates for the evaluations of the Subject in the Academic Year 2017-2018 will be the following:

- January-February 2018
- September 2018

2.Learning goals

2.1.Learning goals

El estudiante, para superar esta asignatura, deberá demostrar los siguientes resultados:

1. - Conocer el concepto y definición de enfermedad rara.
2. - Conocer la prevalencia global de las enfermedades raras en nuestro entorno (España, Europa).
3. - Conocer las generalidades clínicas/terapéuticas de las enfermedades raras.
4. - Conocer los principales aspectos de los Planes y Estrategias (nacional y europea) sobre las enfermedades raras.
5. - Conocer la importancia de las enfermedades raras en Pediatría.
6. - Conocer los principales recursos disponibles para el estudio, investigación y atención socio-sanitaria a los pacientes y familias con enfermedades raras.
7. - Conocer la clínica, diagnóstico y tratamientos de las principales enfermedades raras en la edad pediátrica.

2.2.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.Aims of the course and competences

3.1.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.

3.2.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.

4.Assessment (1st and 2nd call)

26739 - Unusual Children's Illnesses

4.1. 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 (type test): 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:
First Call: January-February 2018
Second Call: September 2018

5. Methodology, learning tasks, syllabus and resources

5.1. Methodological overview

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

1. Lectures (theoretical) 1 hour (13 classes = 13 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. Personal work on some of the rare diseases included in the Syllabus, or any other (general) aspects of Rare Diseases.

5.2. Learning tasks

- Attendance to theoretical lessons
- Assistance and discussion of clinical cases
- Performing personal work related to rare diseases

5.3. Syllabus

The program that the student is offered to achieve the expected results includes the following activities (Theoretical and practical -clinical case- aspects):

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

5.4. Course planning and calendar

26739 - Unusual Children's Illnesses

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.

Scheduled (tentative) dates for evaluations:

- January/February, 2018
- September 2018

5.5. Bibliography and recommended resources

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