

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

Code	36355
Name	Enfermedades raras
Cycle	Grade
ECTS Credits	4.5
Academic year	2019 - 2020

Study (s)

Degree	Center	Acad. year	Period
1204 - Degree in Medicine	Faculty of Medicine and Odontology	4	Second term

Subject-matter

Degree	Subject-matter	Character
1204 - Degree in Medicine	18 - Optional subjects	Optional

Coordination

Name	Department
CODOÑER FRANCH, PILAR	290 - Pediatrics, Obstetrics and Gynaecology

SUMMARY

The main goal in this optional subject 'Rare Diseases' is to create an academic and formative space, which provides students with tools for their future professional development, when facing an orphan disease. This subject intends to supply the lack of specific formation in this field, with special attention to the fact of addressing rare diseases in the learning process which students experience, as specific knowledge and skills are required. A formative strategy which comprises specific content regarding rare diseases is applied. It also considers measures the main of objective of which is to promote interest and sensitivity towards the problems these diseases cause.

PREVIOUS KNOWLEDGE



Relationship to other subjects of the same degree

There are no specified enrollment restrictions with other subjects of the curriculum.

Other requirements

Any degree studies would be enough to access epidemiological knowledge. However, their application in the field of rare diseases requires that students are familiar with aspects of biology and human physiopathology.

OUTCOMES

1204 - Degree in Medicine

- Students must be able to apply their knowledge to their work or vocation in a professional manner and have acquired the competences required for the preparation and defence of arguments and for problem solving in their field of study.
- Students must have the ability to gather and interpret relevant data (usually in their field of study) to make judgements that take relevant social, scientific or ethical issues into consideration.
- Understand and recognise the effects, mechanisms and manifestations of diseases over the structure and function of the human body.
- Recognise health determinants in population, such as genetic ones, dependent on sex, lifestyle, demographic, environmental, social, economic, psychological and cultural.
- Proper organisation and planning of the workload and timing in professional activities.
- Team-working skills and engaging with other people in the same line of work or different.
- Criticism and self-criticism skills.
- Capacity for communicating with professional circles from other domains.
- Acknowledge diversity and multiculturality.
- Consideration of ethics as a fundamental value in the professional practise.
- Working capacity to function in an international context.

LEARNING OUTCOMES

1. Students will acquire knowledge about:

- a. The notion of rare disease.
- b. Different clinical aspects related to rare diseases.



- c. Localisation of resources and information about them.
- d. Aspects of prevention and early detection, and the techniques used for it: neonatal screening, genetic diagnosis, etc.
- e. Therapeutical aspects: advanced therapies, orphan drugs, contributing factors and healthcare products.
- f. Aspects regarding social issues and health care.
- g. New lines of research in the field.

2. Students will know the most relevant aspects of:

- a. General and specific characteristics of rare diseases.
- b. General and specific characteristics of orphan drugs.
- c. National and international legal framework, and policies applied within the field of rare diseases and orphan drugs.
- d. Main organisations, institutions, health portals and networks related to rare diseases.

3. Students will develop competencies of variable size in relation to the following:

- a. Associations of patients suffering from rare diseases.
- b. Coordination between levels of health care. Pharmaceutical care and pharmacotherapeutical follow-up in the field of rare diseases.

DESCRIPTION OF CONTENTS

1. THEORETICAL THEMES.

1. Rare diseases. Introduction and general aspects. Epidemiology. General classification of rare diseases.
2. Bases of heredity and patterns of Mendelian Inheritance. Knowledge of the non-Mendelian models of Inheritance (mitochondrial, sex-linked, genomic imprinting, etc.). Genetic polymorphisms and mutations. The concept of inherited disease (penetrance, expressivity, genetic and phenotypic heterogeneity).
3. Molecular bases of rare diseases. Diseases due to a defect in the synthesis or catabolism of complex molecules: lysosomal and peroxisomal diseases, diseases of the intracellular transportation and processing. Diseases due to accumulation of toxic substances: aminoacidopathies, organic aciduria, disorders of the urea cycle, sugar intolerance. Diseases due to energy-deficit, such as glycogen storage disease, defects in the gluconeogenesis, congenital lactic acidemia, oxidation disorders of fatty acids, and diseases of the mitochondrial respiratory chain.
4. Strategy for Rare Diseases in the National Health Care System (health records, early detection and diagnosis). Rare diseases legislation. The importance of biomedical research on rare diseases.



Information tools on rare diseases (Orphanet, Eurodis).

5. Congenital anomalies.

6. Parasitic and infectious rare diseases.

7. Nephro-urological rare diseases.

8. Endocrine and metabolic rare diseases.

9. Rare diseases of the nervous system and sense organs.

10. Rare diseases of the cardiovascular system.

11. Rare diseases of the respiratory system.

12. Rare diseases of the osteo-myoarticular system and the connective tissue.

13. Rare onco-haematological diseases.

14. Rare diseases of immunity origin. Autoimmune diseases.

15. Rare diseases of skin and subcutaneous tissue.

16. Psychosocial aspects of RD.

17. Therapeutical aspects of RD. The concept of orphan drugs.

2. PRACTICAL LESSONS

1. There will be workshops (in the computer room) on the different clinical disciplines which teach students the skills needed to suspect diseases, deal with patients and their environment, and provide them with everything which is essential to mitigate a disease (pharmacological and non-pharmacological treatments, rehabilitation, social-sanitary support, etc.).

2. Presentation of clinical cases as seminars.

WORKLOAD

ACTIVITY	Hours	% To be attended
Theory classes	19,00	100
Seminars	16,00	100
Computer classroom practice	10,00	100
Attendance at events and external activities	10,00	0
Development of group work	8,00	0
Development of individual work	10,00	0
Study and independent work	6,00	0
Readings supplementary material	4,00	0
Preparation of evaluation activities	6,00	0
Preparing lectures	8,00	0
Preparation of practical classes and problem	8,00	0
Resolution of case studies	4,00	0
Resolution of online questionnaires	3,50	0
TOTAL	112,50	



TEACHING METHODOLOGY

Regarding theoretical credits, professors will present the most important methods, procedures and content through master classes, in order to help students develop several skills and meet the objectives. Students' participation in the class should be encouraged and they will have access to the didactic material professors may have used through the electronic platform *Aula Virtual*.

Practise in the classroom: seminars. In small groups, professors will present deeply specialised themes, case studies, management of bibliography, current issues... Team work is encouraged, as well as oral presentation skills, which could be interpreted as 'cooperative learning'.

Clinical case studies. Intensive and complete analysis of a fact, a problem or a real situation in order to know about it, how to interpret it, contrast data, make a diagnosis and, in some occasions, be trained on how to apply possible alternative solutions.

Practise in Units of Paediatric Dysmorphology, Medical Genetic Services and Associations of patients suffering from rare diseases.

EVALUATION

The evaluation will be continuous and all formative activities taken will be considered.

As regards to the theoretical evaluation (50% of the final score), students will take a test with multiple-choice questions in which incorrect answers will not penalise (40% of the score), plus attendance to the different companies, entities or institutions (10% of the score).

As regards to the practical evaluation (50% of the final score), students will do exercises, both individually and in groups, with oral presentation work and defence of their own assignments (40% of the score). Attendance to the different companies, entities or institutions will also be taken into account (10% of the score).

Students can pass the subject if they obtain 5 as a mark or superior – having a minimum of 2.5 in the theoretical part and 2.5 in the practical one.

In this subject, students will not be allowed to write their test (or even take it before the agreed date) if they have not completed their training (internship).

Attendance of practices will be compulsory.



REFERENCES

Basic

- IZQUIERDO M, AVELLANEDA A. Enfermedades Raras: Un enfoque práctico. 1a Edición. Madrid: ISCIII; 2004.
- Servicio de Pediatría. Hospital Universitario «Marqués de Valdecila». Universidad de Cantabria. Necesidades de los pacientes pediátricos con enfermedades raras y de sus familias en Cantabria. Documentos 69/2005. Real Patronato sobre Discapacidad. Ministerio de Trabajo y Asuntos Sociales.
- POSADA DE LA PAZ M, GROFT STEPHEN C, eds. Rare diseases epidemiology. Series: Advances in Experimental Medicine and Biology, Vol. 686. 1ªed. Springer; 2010.
- Instituto de Investigación en Enfermedades Raras. ISCIII. Registro de Enfermedades Raras. <https://registoraras.isciii.es>
- KLEIN DE ZIGHELBOIM E, GALLARDO JUGO BE, CHÁVEZ PASTOR M, ABARCA BARRIGA HH. Atlas de Dismorfología Pediátrica. Fondo Editorial del Instituto Nacional de Salud del Niño. Lima, Perú 2012.
- Estrategia en enfermedades raras del Sistema nacional de Salud. Sanidad 2009. Ministerio de Sanidad y Política Social.

ADDENDUM COVID-19

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

1. Contenidos

Se mantienen los contenidos inicialmente recogidos en la guía docente, excepto la reunión con la Asociación de enfermos afectados del Síndrome de Prader Willi, en la que venían a explicar sus vivencias con la enfermedad. Tenía 2h de duración y se sustituye por trabajo autónomo de los alumnos con la realización de un proyecto de enfoque diagnóstico y terapéutico de un paciente sospechoso de padecer una enfermedad rara.

Se ha mantenido las sesiones programadas en las mismas fechas y horas con la misma duración que tenían en el momento del inicio de la docencia no presencial. Se han ido realizando mediante docencia no presencial las 6 horas que restaban de teoría. Se mantiene las dos horas de sesiones prácticas en las que se realizaba una reunión con Asociaciones de enfermos, pero se sustituyen estas reuniones por un proyecto elaborado por los alumnos mediante trabajo autónomo, con material aportado por los profesores y subido al aula virtual.

3. Metodología docente



Se dispondrá en el aula virtual de las presentaciones de las clases y de los seminarios. Mismos materiales previstos en la guía docente original para la docencia presencial. Utilización del foro del aula virtual para aclarar dudas. Se propondrá la realización de un proyecto elaborado por los alumnos acerca del enfoque diagnóstico y terapéutico de un paciente con enfermedad rara, que será realizado en grupos de 4-5 alumnos. Cada uno especificará la tarea que ha realizado en el grupo. Se proporcionará bibliografía en el aula virtual.

Se mantiene la programación de tutorías virtuales (atención en 48 h laborables máximo por correo electrónico).

4. Evaluación

Se suprime la evaluación teórica y se pasa a un sistema de evaluación práctica del trabajo realizado por los alumnos en la que :Se mantienen las notas resultantes de la evaluación continua obtenidas antes de la entrada en vigor del estado de alarma (50 % de la calificación final).

Elaboración de un proyecto de enfoque diagnóstico y terapéutico de un paciente con enfermedad rara que será elaborado de forma grupal (4-5 alumnos) en el que se determinará el papel y las tareas a realizar por cada uno de los integrantes del grupo. Se subirá al aula virtual, en el apartado Tarea y se enviará por "Quickmail" del aula virtual (50 % de la calificación final).

5. Bibliografía

La bibliografía recomendada se mantiene y se añadirá textos informativos para la realización del proyecto como lecturas recomendadas, que se subirán como material al aula virtual por parte del profesorado.