26739 - Unusual Children's Illnesses

Información del Plan Docente

Academic Year	2016/17
Academic center	104 - Facultad de Medicina
Degree	304 - Degree in Medicine
ECTS	4.0
Course	5
Period	First semester
Subject Type	Optional
Module	---

1.Basic info

1.1.Recommendations to take this course

1.2.Activities and key dates for the course

2.Initiation

2.1.Learning outcomes that define the subject

2.2.Introduction

3.Context and competences

3.1.Goals

3.2.Context and meaning of the subject in the degree

3.3.Competences

3.4.Importance of learning outcomes

4.Evaluation

5.Activities and resources

5.1.General methodological presentation

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.

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5.2.Learning activities

5.3.Program

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. Bone dysplasias: Achondroplasia
Lesson 7. Inborn Errors of Metabolism (IEM)
Lesson 8. Spinal Muscular Atrophy type 1 (Werdnig- Hoffmann Disease)
Lesson 9. Prader-Willi Syndrome
Lesson 10. Cornelia de Lange Syndrome
Lesson 11. Myotonic Dystrophy (Steinert's Disease)
Lesson 12. Mitochondrial Diseases

5.4.Planning and scheduling

Scheduled lectures and presentation of works:

The lectures (theoretical + clinical case) will be held from 17:00 to 19:00h on the laborable Mondays during the 9th semester of the Grade. Should be festive monday, an alternative weekday will be sought.
Scheduled (tentative) dates for evaluations:
- February 1, 2017
- September 6, 2017

5.5.Bibliography and recomended resources

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- o Darras BT. Spinal Muscular Atrophies. En: Pediatr Clin North Am 2015; 62(3):743-766. Epub 2015 Apr 11 [PUBLICACIÓN PERIÓDICA]
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- o Danielsson K, Mun LJ, Lordemann A, Mao J, Lin CH. Next-generation sequencing applied to rare diseases genomics. Expert Rev Mol Diagn 2014; 14(4):469-487. Epub 2014 Apr 4. [PUBLICACIÓN PERIÓDICA]
- o Cassidy SB, Schwartz S, Miller JL, Driscoll DJ. Prader-Willi syndrome. Genet Med 2012; 14(1):1026. [PUBLICACIÓN PERIÓDICA]
- o McLennan Y, Polussa J, Tassone F, Hagerman R. Fragile X Syndrome. Curr Genomics 2011; 12:216-224. [PUBLICACIÓN PERIÓDICA]
- o Aymé S. The importance of review articles in making the voice of rare diseases Heard. OJRD 10th Anniversary. Orphanet J Rare Dis 2016 28;11(1):71. doi: 10.1186/s13023-016-0456-5. No abstract available.

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- Evangelista T, Hedley V, Atalaia A, Johnson M, Lynn S, Le Cam Y, Bushby K. The context for the thematic grouping of rare diseases to facilitate the establishment of European Reference Networks. Orphanet J Rare Dis 2016; 4;11-17. doi: 10.1186/s13023-016-0398-y.

- Paz S, Torrent J, Poveda JL, Perez J, Moreno JL, Martin A, Gonzalez L, Cruz J, Comellas M, Abaitua I, Urcelay J. Experts consensus on the future of Rare Diseases care and orphan drugs access In Spain: A Delphi Study. Value Health 2015; 18(7):A679. doi: 10.1016/j.jval.2015.09.2016. Epub 2015 Oct 20. PMID: 26533805.