Universitat Autònoma de Barcelona

2022/2023

Medical Genetics

Code: 102886
ECTS Credits: 3

Degree	Type	Year	Semester
2502442 Medicine	OT	3	2
2502442 Medicine	OT	4	2
2502442 Medicine	OT	5	0
2502442 Medicine	OT	6	0

Contact

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Use of Languages

Principal working language: catalan (cat)
Some groups entirely in English: No
Some groups entirely in Catalan: No
Some groups entirely in Spanish: No

Teachers

Alberto Plaja Rustein

Prerequisites

It is advisable that students have a good knowledge of English because many of the sources of information on this subject are in this language. It would have been better if the Competences corresponding to the subjects: Cell Biology and Human Genetics.

Objectives and Contextualisation

The main objectives of this subject are: Know the genetic basis of the main diseases with a genetic basis or component. Relate genetic dysfunction with the pathological phenotype. Perform the genetic interpretation of diagnosis, prognosis, prevention and therapy of pathologies Genetics more frequent in the human population. Know the distribution of genetic-based diseases in a given population taking into account its origin. Analyze from a genetic point of view the probandus-family relationship that facilitates the offer of advice genetic.

Competences

Medicine

- Demonstrate a sufficient command of English, both oral and written, for effective scientific and professional communication.
- Demonstrate an understanding of the fundamentals of action, indications, efficacy and benefit-risk ratio of therapeutic interventions based on the available scientific evidence.
- Demonstrate basic research skills.

- Demonstrate understanding of the importance and the limitations of scientific thought to the study, prevention and management of diseases.
- Demonstrate understanding of the mechanisms of alterations to the structure and function of the systems of the organism in illness.
- Demonstrate understanding of the organisation and functions of the genome, the mechanisms of transmission and expression of genetic information and the molecular and cellular bases of genetic analysis.
- Demonstrate, in professional activity, a perspective that is critical, creative and research-oriented.
- Indicate the basic diagnosis techniques and procedures and analyse and interpret the results so as to better pinpoint the nature of the problems.

Learning Outcomes

1. Apply the basic principles of the scientific method (observation of phenomena, hypothesis formulation and testing of hypotheses) to the diagnosis, treatment and prevention of human diseases.
2. Demonstrate a sufficient command of English, both oral and written, for effective scientific and professional communication.
3. Demonstrate basic research skills.
4. Demonstrate, in professional activity, a perspective that is critical, creative and research-oriented.
5. Describe the diagnosis, prognosis, prevention and treatment for the most common genetic pathologies in the human population.
6. Describe the molecular basis of the mechanisms underlying anatomopathological alterations of various diseases, primarily neoplastic and hereditary ones, in different body systems.
7. Identify the molecular basis of the main genetic diseases with a biochemical translation.
8. Identify the most efficient molecular biology tests for prevention, diagnosis and control of treatment for the most common human pathologies.
9. Interpret research results and their application to clinical practice.

Content

Introductory session (in person)

Follow-up sessions of the subject .
Final session of presentation and discussion of the work done by the stu
Electronic modules
Module 1. Most frequent monogenic diseases
Module 2. Metabolic diseases
Module 3. Mental and behavioral diseases
Module 4. Endocrine diseases
Module 5. Hematological diseases
Module 6.

Microdeletion and gene microduplication syndromes

Methodology

Directed: The course includes 2 face-to-face sessions, one at the beginning and the other at the end of the course. Supervised: Resources and tools available will also be made available, potentially useful for the development and maximum use of the course. The student acquires the basic scientific knowledge of The subject. The student will be able to find the material utlititzado in class in the Virtual Campus. Autonomous: The course includes an electronic syllabus consisting of 6 modules, in which the theoretical content of the course, which will complement the personal study of the topics discussed.

Annotation: Within the schedule set by the centre or degree programme, 15 minutes of one class will be reserved for students to evaluate their lecturers and their courses or modules through questionnaires.

Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
THEORY (TE)	4	0.16	2, 5
Type: Supervised			
VIRTUAL CLASSES (VIRT)	8	0.32	2, 5
Type: Autonomous			
PREPARATION OF WRITTEN WORKS	23	0.92	2, 3, 4
SELF-STUDY	35	1.4	2, 5

Assessment

The competences of this course will be evaluated:

1- Resolution of a clinical case. Represent 10% of the final grade.

2- Realization of a written work on a specific genetic disease. It will represent 50% of the Final mark.

3- Oral presentation of the work done. Represent 40% of the final grade.

Recovery exam: those students who have not passed the continuous assessment will complete a Written exam concerning the contents of the syllabus of the subject.

Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Narrative records/written works	10%	1	0.04	7
Oral defense of works	40%	2	0.08	9
Written evaluation:Objective tests	50%	2	0.08	1, 2, 3, 4, 5, 6, 7, 8, 9

Bibliography

• Bain, Barbara J. *Haemoglobinopathy Diagnosis*. Editorial Blackwell Science, 2001 .(Hi ha la 2a edició de 2006)

• Epstein, Richard J. *Human Molecular Biology*. Editorial Cambridge University Press,2003

• Farreras-Rozman. *Medicina interna*. Editorial Elsevier, Vol, 1 i 2, 2009

• Firth, Helen V. i Hurst, Jane A. *Oxford Desk Reference Clinical Genetics*. Editorial Oxford University Press, 2005

• Gardner, R. J. McKinlay i Sutherland, Grant R. *Chromosome abnormalities and Genetic Counseling*, 3a ed. Oxford Monographs on Medical Genetics. Editorial Oxford University Press, 2011

• Genetics and Genomics in Medecine. Strachan et al. (2015). Ed Garland Science, 1rst ed

• Harper, Peter S. *Practical genetic counselling*, 6a ed. Editorial Hodder Arnold, 2004 (Hi ha la 7a edició de 2010)

• Jord- Genetics and Genomics in Medecine. Strachan et al. (2015). Ed Garland Science, 1rst ede, L. B. et al. Genética Médica, 4a ed. Elsevier, 2011

• Korf B.R i Irons M.B. Human Genetics and Genomics (4ª ed) Wiley-Blackwell, 2013

• Klug V.S. et al. Conceptos de Genética (10ª ed) Pearson, 2013

• Nussbaum, Robert L. *Thompson and Thompson Genética en Medicina*, 7a ed. Editorial Masson, 2008

• Read, A. i Donnai, D. New Clinical Genetics. Editorial Scion Publishing Ltd, 2011

• Rimon, David L. *Emery and Rimoin's principles and practice of medical genetics*, 4a ed. Editorial Churchill Livingstone. Vol 2-3, 2002 (Hi ha la 5a edició de 2006)

• Salvador, Joaquín i Carrera, José M. Síndromes congénitos malformativos.Colección de Medicina Materno-Fetal. Editorial Masson, 1995

• Scriver, Charles R. et al. *The Metabolic &molecular bases of inherited disease*, 8th. ed. Editorial MacGraw-Hill, New York, 2001

• Stracher, T. et al. Genetics and Genomics in Medicine, 1st ed. Garland Science, 2014 • Wright , A. i Hastie, N. G

• Wright , A. i Hastie, N. *Genes and common diseases: Genetics in Modern Medicine*. Editorial Cambridge University Press, 2007

Software

The application software is the set of PROGRAMS that allow document management, data processing, digital retouching of images, Internet browsing ... etc. The application software can be grouped into the following sections:
• Word processors: allow the creation of documents with the integration of text, data and images. Examples: Word, WordPad ...
• Databases: they allow the processing of large amounts of information and facilitate subsequent consultation. Example: Access.
• Spreadsheets: allow the processing of data, but in this case numerical, and the performance of mathematical calculations. Example: Excel.
• Communication programs: allow communication through computer networks. You can browse the Internet, send an email ... Examples: IE, Browser, Messenger, Outlook, Eudora ...

• Graphic design: they allow the realization and digital treatment of drawings, plans and photographs. Examples: AutoSketch (technical drawing), Paint (drawing), Paint Shop Pro (image processing) ...
• Multimedia creation programs: allow you to integrate text, graphics, sound and animations, and create interactive documents. Examples: Flash, PowerPoint ...
• ... and the list of programs could continue with file compressors, MP3 players ...
Diagnostic software is the set of programs that allow you to configure and check the correct operation of all the hardware elements of a computer system.