Universitat Autònoma de Barcelona

2017/2018

Medical Genetics

Code: 101886
ECTS Credits: 6

Degree	Type	Year	Semester
2501230 Biomedical Sciences	OB	3	2

Contact

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Use of languages

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

Teachers

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Prerequisites

It would be appropriate to have passed and achieved the competences corresponding to the subjects: Cell Biology, Human Genetics, Molecular Cell Biology, Developmental Biology and Teratogeny.

Objectives and Contextualisation

The main objectives are:

Know the genetic basis of the main diseases with a base or genetic component.
Relate the genetic dysfunction with the pathological phenotype.
Perform the genetic interpretation of the diagnosis, prognosis, prevention and therapy of the most frequent genetic pathologies in the human population.
Understand the distribution of genetic-based diseases in a population taking into account their origin.
Analyze genetically the probands-family relationship that facilitates the offert of a genetic counseling.

Content

1. General principles

1.1. Basic introduction to genetic diseases

1.2. Predisposition or genetic susceptibility

1.3. Genetic counseling

2. Chromosomal alterations

2.1. Autosomal trisomies

2.2. Alterations of sex chromosomes

2.3. Chromosome Microdeletions and Microduplications

3. Neuromuscular diseases

3.1. Muscular dystrophies: definition and classification

3.2. Muscular dystrophies of Duchenne and Becker

3.3. Other muscular dystrophies

3.4. Myotonic dystrophy

3.5. Spinal muscular atrophy

3.6. Charcot-Marie-Tooth's Disease: Sensoriomotor Hereditary Neuropathy

4. Mental and behavior diseases

4.1. Intellectual Disability

4.2. Fragile X syndrome and associated diseases

4.3. Rett syndrome

4.4. Attention deficit disorder and hyperactivity disorder

4.5. Autism

4.6. Schizophrenia

5. Diseases of the central nervous system

5.1. Huntington's disease

5.2. Alzheimer's disease

5.3. Parkinson's

5.4. Primary tumors of the central nervous system

6. Skeletal diseases and connective tissue

6.1. Imperfect osteogenesis

6.2. Acondroplasia

6.3. Marfan Syndrome

6.4. Sarcoma

7.1. Craniosynostosis

8. Dermatological diseases

8.1. Albinism

8.2. Skin cancer

9. Ophthalmological diseases and deafness

9.1. Defects of vision of colors

9.2. Deafness

10. Cardiovascular diseases

10.1. Hypertension

11. Gastrointestinal diseases

11.1. Celiaquia

11.2. Colon cancer

12. Respiratory diseases

12.1. Lung cancer

13. Metabolic diseases

13.1. Alterations in the metabolism of the glucose: lactose intolerance

13.2. Alterations in the metabolism of lipids: Hypercholesterolemia

13.3. Alterations in the metabolism of amino acids: Phenylketonuria

14. Hematological diseases

14.1. Hemoglobinopathies

14.2. Haemophilia

14.3. Leukemia and Lymphoma

15. Urogenital diseases

15.1. Policystics kidney

15.2. Kidney cancer

15.3. Bladder cancer

15.4. Prostate cancer

16. Endocrinological diseases

16.1. Diabetes mellitus

16.2. Breast cancer