

Medical Genetics

Code: 101970
ECTS Credits: 6

Degree	Type	Year	Semester
2500890 Genetics	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, Cytogenetics and Reproductive Genetics.

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 offer 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: Sensorimotor 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. Celiac disease
 - 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. Hemophilia
 - 14.3. Leukemia and Lymphoma
- 15. Urogenital diseases
 - 15.1. Polycystic kidney
 - 15.2. Kidney cancer
 - 15.3. Bladder cancer
 - 15.4. Prostate cancer
- 16. Endocrinological diseases
 - 16.1. Diabetes mellitus
 - 16.2. Breast cancer