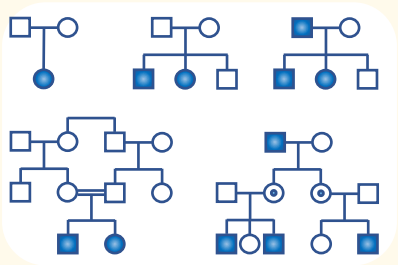

Molecular GeneticS of Human Diseases

Béatrice PARFAIT – Djihad HADJADJ - Eric PASMANT

Effectives: 35
Language: English
Prerequisites:
Sciences/Health studies

Where?
Faculté de Pharmacie
4, avenue de l'Observatoire
75006 PARIS

When
28/11 to 2/12/2022

Evalutation:
Written examination with article analysis

Questions:
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Number ECTS: 3

Total numbers of hours:
30h

Teaching format:
conferences

Teaching objectives

The aim is to provide solid knowledge and skills to answer to the questions and challenges of Genomic Medicine and Human Molecular Genetics: what are the molecular bases of human diseases? How disease-associated genes are identified and analyzed? How the function of these genes can be investigated? How this knowledge can be used to develop therapeutics approaches?

Courses are performed by specialists from reputable research institutes in fundamental and translational research in the field of Human Genetics

Principles of Genomic Medicine based on the new capabilities of exploring the entire human genome in search of the genetic molecular basis of hereditary diseases and developmental anomalies in human, functional study strategies, *in vitro/vivo* models and therapeutics approaches will be presented

Teaching outline

Mutations: Types, mechanisms and functional consequences / Non-coding RNAs and conserved non-coding sequence in hereditary diseases / Epigenetic abnormalities / Animal models / Pharmacological therapies of hereditary diseases

Molecular bases and therapeutic strategies in: hereditary kidney diseases / Krebs cycle alterations in cancer / Mitochondrial diseases / Childhood-onset retinal blindness / Microsatellite instability and trinucleotide repeat expansion disorders / Facio-Scapulo-Humeral dystrophy / Spinal Muscular Atrophy / Duchenne Muscular Dystrophy