

Université Paris Cité - Sciences du Vivant – Master de Génétique – M2

Mention européenne de GENETIQUE

UE Génétique moléculaire des maladies génétiques

Molecular Genetics of Human Diseases

Responsables : Béatrice Parfait, Eric Pasmant et Jihad Hadjadj

Faculté de Pharmacie de Paris
4, avenue de l'Observatoire – 75006 Paris

Salle Géranium

Lundi

10h30-12h30

Mutations: Types, Generative Mechanisms and functional Consequences

Jian-Min CHEN

UMR 1078, EFS, UBO, Brest

13h30-16h30

Human Genetics, Evolution and Disease: an overview

Richard REDON

Institut du thorax, Inserm UMR 1087 / CNRS UMR 6291, Faculté de Médecine, Nantes

Mardi

9h15-10h45

Molecular genetics of neurocristopathies

Nadège BONDURAND

UMR 1163, Institut Imagine, Hôpital Necker, Paris

11h00-13h00

Subtelomeric imbalance: the exemple of facio-Scapulo-Humeral dystrophy

Frédérique MAGDINIER

U910 INSERM, Faculté de Médecine, Marseille

14h00-15h30

Molecular bases of hereditary kidney diseases

Sophie SAUNIER

UMR 1163, Institut Imagine, Hôpital Necker, Paris

15h45-17h15

Animal models of human hereditary diseases

Carine LE GOFF

U1148 INSERM, Hôpital Bichat, Paris

Mercredi

11h15-12h45

Epigenetic abnormalities

Laila EL KHATTABI

Institut Cochin, UMR INSERM 1016, Paris

14h00-15h30

Conserved non coding sequence of the genome and hereditary diseases

Chris GORDON

UMR 1163, Institut Imagine, Hôpital Necker, Paris

16h00-17h30

Molecular genetics of Krebs cycle alterations in cancer

Judith FAVIER

UMR 970 – PARCC Hôpital Européen George Pompidou, Paris

Jeudi

9h00-11h00

Microsatellite instability and trinucleotide repeat expansion disorders.

Mario GOMES-PEREIRA

Inserm UMRS974, Centre de Recherche en Myologie, GH Pitié-Salpêtrière, Paris

11h15-12h45

Molecular genetics of mitochondrial diseases

Agnès RÖTIG

UMR 1163, Institut Imagine, Hôpital Necker, Paris

13h45-15h15

Genetics and potential therapies of childhood-onset retinal blindness

Jean-Michel ROZET

UMR 1163, Institut Imagine, Hôpital Necker, Paris

15h30-17h00

Genomic rearrangements

Thomas COURTIN

CRICM UPMC/INSERM UMRS975/CNRS UMR7225, GH Pitié-Salpêtrière, Paris

Vendredi

9h30-11h00

Pharmacological therapies of hereditary diseases

Arnold MUNNICH

UMR 1163, Institut Imagine, Hôpital Necker, Paris

11h15-12h45

Spinal Muscular Atrophy: Molecular Genetics and Therapeutic Strategies

Alexandra MARTINS

U1079 INSERM, Faculté de Médecine et de Pharmacie, Rouen

13h45-15h15

Update of therapeutic approaches for the Duchenne Muscular Dystrophy: from exon skipping to microdystrophin transfer

Sestina FALCONE

UM76 -UPMC/ U974 – Inserm / UMR7215 - CNRS, Hôpital Pitié-Salpêtrière, Paris

16h00-18h00

Examen sous forme d'analyse d'un article en anglais (tous les documents sont autorisés)