COLLÈGE DE FRANCE
1530

CHAIRE DE MÉDECINE EXPÉRIMENTALE
Alain FISCHER, Professeur

CHAIRE DE GÉNÉTIQUE HUMAINE
Jean-Louis MANDEL, Professeur

Novel therapies for monogenic diseases

colloque en anglais

Jeudi 16 et vendredi 17 avril 2015
Amphithéâtre Maurice Halbwachs

Thursday, April 16th

09h30 Genome engineering with CRISPR-Cas9 : biology, mechanisms and applications in medicine
Emmanuelle Charpentier, *Helmholtz I., Hannover*

10h15 Gene therapy of primary immunodeficiencies (PID)
Alain Fischer, *Imagine Institute, Paris*

11h00 Genetic Engineering of Human Hematopoiesis for the Treatment of Inherited Diseases and Cancer
Luigi Naldini, *San Raffaele Scientific Institute, Milan*

11h45 **Poster session**
Lunch

14h15 Gene therapy of hemophilias
Amit Nathwani, *University College, London*

15h00 New mutation-specific pharmacotherapies for Cystic fibrosis
Kris de Boeck, *Leuven University Hospital*

15h35 Added value of new genomics technologies in undiagnosed patients who may benefit from known therapies
Judith Melki, INSERM, *U. Paris-Sud, Kremlin-Bicêtre*

15h55 From promising clinical studies to recent clinical trials for the fragile X mental retardation syndrome: the challenge of judging efficacy
Vincent des Portes, *CHU Lyon and CNRS/UCBL L2C2 laboratory*

16h30 Pause and poster viewing

17h00 The discovery and development of olesoxime for spinal muscular atrophy
Rebecca Pruss, *Trophos and Winhover Biomed, Marseille*

17h35 From gene identification to functional analysis and treatment of achondroplasia
Laurence Legeai-Mallet, *Imagine Institute, Paris*

17h55 Inhibition of thromboxane synthase, a new mechanism to increase bone density ?
Valerie Cormier-Daire, *Imagine Institute, Paris*

18h15 **Selected oral communication**
Drug screening on Hutchinson Gilford progeria using pluripotent stem cells reveals new pharmacological modulators of prelamin a farnesylation
Xavier Nissan, *ECS I-Stem, Evry*

18h30 **End of the session**

Friday April 17th

09h00 Novel approaches for treatment of retinal diseases
José-Alain Sahel, *Institut de la Vision, Paris*

09h45 Gene replacement therapy for myotubular myopathy
Anna Buj Bello, *Genethon and INSERM, Evry*

10h15 REPS1 is a novel gene of Neurodegeneration with Brain Iron Accumulation
Arnold Munnich, *Imagine Institute, Paris*

10h35 **Pause**

11h00 Efficient AAV gene therapy in cardiac and neurological murine models for Friedreich ataxia
Hélène Puccio, *IGBMC, Strasbourg*

11h30 **Selected oral communications**

High expression and therapeutic efficiency of systemic delivery of a dual AAV strategy in a murine model for dysferlin deficiencies
William Lostal, *Généthon, Evry*

Gene therapy for Alzheimer's disease : the cholesterol connection
Nathalie Cartier, *Inserm MIRCen CEA, Orsay*

12h00 Can we afford cost of innovative therapies for rare diseases ?
Round table with *Franck Dufour (ARC), Serge Braun (AFM)*

12h45 **Conclusions**

13h00 **End of the meeting**

Amphithéâtre Maurice Halbwachs
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Serge Haroche
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