



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 moves from rare to frequent diseases
Marina Cavazzana, *Imagine I., Paris*
- 11h00** Gene replacement and targeted genome editing of
hematopoietic stem cells 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** Discovery and development of olesoxime for spinal muscular
atrophy therapy
Rebecca Pruss, *Trophos and Winhover Biomed, Marseille*
- 17h35** From gene identification to functional analysis and treatment
of achondroplasia
Laurence Legeai-Mallet, *Imagine I., Paris*
- 17h55** Inhibition of thromboxane synthase, a new mechanism to
increase bone density ?
Valerie Cormier-Daire, *Imagine I., Paris*
- 18h15** **Selected oral comm**
- 18h30** **End of the session**

Friday April 17th

- 09h00** Novel approaches for treatment of retinal diseases
Jose L. Sahel, *I. de la Vision, Paris*
- 09h45** Gene replacement therapy for myotubular myopathy
Anna Buj Bello, *Genethon and INSERM, Evry*
- 10h15** The irony of iron chelation in neurodegeneration with brain
iron accumulation
Arnold Munnich, *Imagine I., Paris*
- 10h35** **Pause**
- 11h00** Efficient AAV gene therapy in cardiac and neurological murine
models for Friedreich ataxia
Hélène Puccio, *IGBMC, Strasbourg*
- 11h30** **2 Short selected talks**
- 12h00** Can we afford cost of innovative therapies for rare diseases ?
Round table with *Franck Dufour, ARC, Christophe Duguet,
AFM*
- 12h45** **Conclusions**
- 13h00** **End of the meeting**

