Program

WEDNESDAY, March 5th, 2014

REGISTRATION OPENS AT 12 AM

14 to 17: AFTERNOON SESSION A:

Stem Cell and Cell Therapy Approaches

Chairwoman: Dr. Marisa Jaconi, CH

- Prof. Giulio Cossu, UK "Cell therapy for muscular dystrophies"
- Prof. Alan Tyndall, CH "Stem Cell Therapies of Autoimmune Diseases"
- Prof. Marc Peschanski, FR "Harnessing pluripotent stem cells derivatives to decipher mechanisms and identify treatments for monogenic diseases"
- Prof. Yann Barrandon, CH "A clonal strategy for safe ex vivo gene therapy of epidermis"
- Sarah Decembrini (Abstract No 20) "Derivation of traceable and transplantable photoreceptors from mouse embryonic stem cells"

17 to 18: POSTER SESSION

18 to 20: Public Opening Ceremony

Welcome

- Dr. Jörg Reinhardt, Novartis, Chairmen of the Board of Directors
  Moderation: Prof. Dr. Susan Gasser, CH

- Dr. David M. Lee, Novartis Institutes for Biomedical Research, CH
- Yann Le Cam, EURORDIS, FR
- Prof. Stephen C. Groft, NIH-ORDR, USA "Global Research Collaborations: Are We Reaching Our Goals for Rare Diseases?"
THURSDAY, MARCH 6TH, 2014

9 to 12: MORNING SESSION B:

Mapping diseases and genome instabilities

Chairman: Prof. Dr. Stylianos Antonarakis, CH

- Prof. Thaddeus Dryja, USA "Special considerations for phase I trials of gene therapies for retinitis pigmentosa and allied retinal degenerations"
- Prof. Nicholas Katsanis, USA "Modelling the Morbid Pediatric Genome"
- Prof. Stanislas Lyonnet, FR "Non-coding genome alterations in rare development anomalies"
- Prof. Alexandre Reymond, CH "Chromatin loops and CNVs: the complex spatial organization of the 16p11.2 locus"
- Dr. Davide Gabellini, IT "FSHD muscular dystrophy provides a molecular understanding of the repetitive (epi)genome"
- Meehan Terrence (Abstract No32) "Informing rare disease mechanisms: informatics for the international mouse phenotyping consortium"

12 to 13: Lunch

12 to 14: Lunch-Symposia: Swiss Research Showcase

13 to 14: POSTER SESSION

14 to 17: AFTERNOON SESSION C:

Pathophysiology and diagnostics

Chairman: Dr. Mike Morris, CH

- Prof. Dennis Lo, HK "Noninvasive prenatal testing using cell-free fetal DNA in maternal plasma"
- Prof. Cécile Janssens, USA "How predictive is our DNA?"
- Prof. Ephrat Levy-Lahad, IL "From Mutation to Pathogenesis in Rare Diseases"
- Dr. Anne Puel, FR "Inborn errors of human IL-17 immunity underlie chronic mucocutaneous candidiasis"
- Prof. Orly Elpeleg, IL "Whole Exome Sequencing in Rare Diseases"
- Sánchez-Jiménez Francisca María (Abstract No34) "Searching for Biomedical Relationships among Genes and Diseases: a great ally for rare diseases"

17 to 19: POSTER SESSION
FRIDAY, MARCH 7Th, 2014

9 to 12: MORNING SESSION D:

Bringing treatments to the clinic

Chairman: Dr. Jordi Surrallés, ES

- Prof. Marc Tardieu, FR "Development of an intra-cerebral gene therapy trial in Sanfilippo disease Type A"
- Prof. Michael Sinnreich, CH "Novel treatment strategies for muscular dystrophy"
- Prof. Lakshminarayan Ranganath, UK "Advances in the treatment of Alkaptonuria: the nitisinone experience"
- Prof. Colin McKerlie, CAN "Systematic large-scale gene function analysis of the mouse genome: An opportunity for new mouse models of rare diseases for research and drug discovery"
- Kostic Corinne (Abstract No29) "Optimization of RPE65-gene transfer using a lentiviral vector for LCA treatment"
- Schäfer Matthias (Abstract No35) "Activation of Nrf2 in keratinocytes causes chloracne (MADISH)-like skin disease in mice"
- Jordi Surrallés, (Abstract No21) "Fanconi anemia: from gene discovery to gene therapy"

12 to 13: Lunch

12 to 14: Lunch-Workshop: Neurocognitive Disorders

13 to 14: POSTER SESSION

14 to 17: AFTERNOON SESSION E:

Degenerative disorders

- Prof. Jose-Alain Sahel, FR "Vision restoration strategies in blinding retinal dystrophies"
- Prof. Robert D. Goldman, USA "Altered Intermediate Filament Networks are the Hallmarks of Many Rare Diseases"
- Prof. Colin L. Stewart, SG "Analyzing Progeria to provide insights into the mechanisms of ageing"
- Prof. Gisele Bonne, FR "Laminopathies of the striated muscle: from gene defects towards pathophysiological mechanisms"
- Giacomini Caterina (Abstract No16) "Abnormal Lamin B1 levels affect Neuronal Viability and Differentiation"
- Waddington Simon (Abstract No27) "Perinatal gene therapy rescues acute neonatal lethal Neuronopathic Gaucher Disease in mice"

17 to 19: POSTER SESSION

Delegates’ dinner/party
SATURDAY, MARCH 8TH, 2014

9 to 12: MORNING SESSION F:

Research and patients

Chairman: Dr Nick Sireau, UK

• Karen Aiach, FR "Lessons learned from a pioneering phase I/II gene therapy trial in Sanfilippo syndrome"
• Dr. Martine Zimmermann, CH "Regulatory frameworks and incentives for development of Orphan Medicinal Products"
• Dr. Nick Sireau, UK "Curing Black Bone Disease: lessons from a major clinical trial"
• Prof. Philippe Gorry, FR "Role of academic research in the discovery of Orphan Drugs"
• Prof. Marshall Summar, USA "Rare Disease Registries Successful Models and Lessons"

Program updated February 20, 2014