3rd international conference
SANFILIPPO SYNDROME
AND RELATED DISEASES
A digital event
November 12-13, 2020

SCIENTIFIC PROGRAM - 12 & 13 November 2020

Online registration: https://fondation-sanfilippo.optionk.com

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Our partners / Nos partenaires:
Thursday 12 November 2020

9:15  Prof. Stylianos Antonarakis, Chairman and Professor of Genetic Medicine Emeritus, University of Medicine, Geneva, Switzerland

Introduction and welcome speech

Session 1: Screening and diagnosis

9:30  Dr. Ana Carolina Brusius-Facchin, Division of Medical Genetics, Hospital de Clinicas of Porto Alegre (HCPA), Brazil

Sensitivity, advantages, limitations and clinical utility of targeted next-generation sequencing panels for the diagnosis of selected lysosomal storage disorders

10:00  Prof. Lucy Raymond, Dept. of Medical Genetics, University of Cambridge, UK

The next generation of children project: Whole genome sequencing for rapid diagnosis of severely ill children in intensive care

10:30  Dr. Francisco del Castillo, Genetic Services, Ramon y Cajal Hospital, Madrid, Spain

Efficiency of NG-based gene panels as first-line screening tests for the quick diagnosis of lysosomal diseases.

Session 2: Early stage and investigational treatments

11:30  Dr. Maria Francisca Coutinho, Research and Development Unit, Human Genetics Dept., National Institute of Health, Porto, Portugal

Targeting stable nucleic acid lipid particle (SNALP)-formulated siRNAs to the brain as a therapeutic approach in lysosomal storage disease-associated neuropathy

12:00  Dr. Brian Bigger, Faculty of Medical and Human Sciences, University of Manchester, UK

Understanding innate immunity in Sanfilippo therapies that may improve behavior

12:30  Dr. Anastasia Henry, Denali Therapeutics, South San Francisco, USA

Brain delivery and activity of an intravenously administered enzyme using a blood brain barrier transport vehicle

Session 3: MPS and related diseases

15:00  Dr. Bettina Cockroft, Sangamo Therapeutics, Richmond, California, USA

The ST-920 program for Fabry disease

16:00  Dr. Mireille Tallandier, Inventiva Pharma, Daix, France

IMProveS: a Phase Ila clinical trial of odiparcil for the treatment of adults with MPS VI
Friday 13 November 2020

- **Session 4 Clinical studies in MPS III: gene therapy**

  9:30 Dr. Simon Jones, Willink Unit, Manchester Centre for Genomic Medicine, St Mary Hospital, Manchester Academic Health Sciences Centre, University of Manchester, UK  
  Clinical trial design for treatment of Mucopolysaccharidosis Type III

  10:00 Ms. Samantha Parker, Lysogene, Paris, France  
  Lysogene gene therapy program for MPS IIIA

  10:30 Dr. Juan Ruiz, Abeona Therapeutics, Dallas, Texas, USA & Dr. Maria José de Castro, Hospital Clínico Universitario de Santiago de Compostella, Spain  
  Abeona’s gene therapy programs for MPS IIIA and MPS IIIB

  11:00 Prof. Rob Wynn, Blood and Marrow Transplant Unit, Royal Manchester Children’s Hospital and University of Manchester, UK  
  Report of the first ever lentiviral vector, gene-modified stem cell transplant in a patient with MPS IIIA

- **Session 5: Clinical studies in MPS III: ERT, SRT and small molecules**

  13:00 Dr. Steve Maricich, Allievex Corporation, Marblehead, Massachusetts, USA  
  Allievex intracerebroventricular enzyme replacement therapy with tralesinidase alfa for treatment of MPS IIIB

  14:30 Dr. Raj Mehra, Seelos Therapeutics, New York, USA  
  Trehalose (SLS-005) a small molecule for MPS III, OPMD and SCA3

- **Session 6: Round table Q & A**

  15:30 Session with scientific experts (90mn) - Open to all registered participants  
  Chair: Dr. Armand Bottani, Valais Hospital, Central Hospital Institute, Medical Genetics Services, Sion, Switzerland

  17:00 Closing speech