2nd Conference on Sanfilippo Syndrome and Related Diseases
26-28th November 2015, Starling Hotel, Geneva, Switzerland

Day 1 Afternoon
15:00-17:00: Registration and Mixer
17:00-17:30: Welcome speeches
17:30-18:15: Plenary lecture
18:30: Cocktail

Day 2 Morning
8:00-10:00 Session 1: LSD Screening and Diagnosis

1) Keynote presentation
Prof. John Hopwood, Lysosomal Diseases Research Unit, South Australian Health and Medical Research Institute, Adelaide, Australia

2) Newborn screening and selective biochemical screening
Dr. Zoltan Lukacs, Institute of Clinical Chemistry/Dept. of Pediatrics, University Medical Center Hamburg-Eppendorf, Germany

3) Molecular Analysis
Dr. Andreas Gal, Institute of Human Genetics, University Medical Center Hamburg-Eppendorf, Germany

4) Diagnostic challenges in Niemann Pick C
Dr. Heiko Runz, Dept. Genetics and Pharmacogenomics, Merck Research Labs, Boston, USA

10:00-10:30 Coffee break

10:30-12:00 Session 2: Treatment options

1) ERT and substrate reduction
Dr. Eugen Mengel, Center for Pediatric and Adolescent Medicine, University Medical Center, Mainz, Germany

2) Stem cell transplantation
Dr. Rob Wynn, Blood and Marrow Transplant Unit, Royal Manchester Children’s Hospital, and University of Manchester, UK
3) Virus-based delivery systems for human gene therapy
Dr. Alessandra Biffi, San Raffaele Telethon Institute for Gene Therapy (HSR-TIGET), Hospedale San Raffaele, Milan, Italy

12:00-13:30 Lunch

Day 2 Afternoon
13:30-15:00 Session 3: Specific clinical aspects of MPS III and new treatment approaches

1) Keynote presentation - Intra-cerebral administration of AAV2/5 vector containing human NAGLU cDNA in children with MPSIIIB: results at 12 months of a Phase I/II trial
Prof. Marc Tardieu, Paediatric neurology service, INSERM unit 1012, Université Paris South, France

2) Natural history
Prof. Frits Wijburg, Dept. of Paediatrics, Academic Medical Centre, Amsterdam, The Netherlands

3) Antibody-directed, receptor-mediated transcytosis technology to treat MPS IIIa and other LSDs
Dr. James Callaway, ArmaGen Inc. Calabasas, CA, USA

4) Gene therapy and other clinical approaches for MPSIIIC
Dr. Brian Bigger, Faculty of Medical and Human Sciences, University of Manchester, UK

15:30-16:00 Coffee break

16:00-18:00 Session 4: Clinical trials in MPS and related LSDs

1) Keynote presentation - Use of biomarkers in evaluating response to therapy in clinical trials for LSDs
Prof. Hans Aerts, Department of Biochemistry, Leiden University, The Netherlands

2) Gene therapy for MPS IIIA – the P1-SAF-301 trial
Ms. Samantha Parker, Lysogene, Neuilly-sur-Seine, France

3) Planned study of ABX-A and ABX-B for MPS Type A and B
Dr. Tim Miller, Abeona therapeutics, USA

4) Planned clinical studies of non-competitive pharmacological chaperones for GM1-gangliosidosis
Dr. Marc Martinell, Minoryx therapeutics, Barcelona, Spain
18.00-19.00 Poster viewing

**Day 3 morning**

8:30-10:30 Session 5: Regulations

1) The drug development process in rare disease research  
**Dr. Stéphane Demotz, Dorphan SA, Lausanne, Switzerland**

2) The role of patient organisations in shaping rare disease policy  
**Ms. Paloma Tejada, EURORDIS, Paris France**

3) Patient registries in drug development and regulatory decisions for LSDs  
**Dr. Simon Jones, Willink Unit, Manchester Centre for Genomic Medicine  
St Mary's Hospital, Manchester Academic Health Sciences Centre, University of Manchester, UK**

4) Pediatric Perspectives in Drug Development: Switzerland  
**Prof. Dr. Hulya Ozsahin, MD, Exec. MBA Healthcare, Swissmedic-Swiss Agency for Therapeutic Products, Bern, Switzerland**

10:30-11:00 Coffee break

11:00-13:00 Session 6: Families

1) Genetic counseling and prenatal diagnosis (20 min)  
**Dr. Armand Bottani, Dept. Medical Genetics, Geneva University Medical Center, Switzerland**

2) Main highlights of the scientific conference (45 min)

3) Round table Q&A session with scientific experts (40 min)  
**Chair: Dr. Armand Bottani**

**Participants to be confirmed**

12:45-14:15 Lunch  
Departure