

**2nd Conference on Sanfilippo Syndrome and Related Diseases  
26-28<sup>th</sup> 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