

## 4TH INTERNATIONAL CONFERENCE ON SANFILIPPO SYNDROME AND RELATED DISEASES

### DAY 1 – THURSDAY, NOVEMBER 13, 2025

**09:15**

Introduction and welcome

**Stylios Antonorakis**

#### Gene Therapy and ERT

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**09:30 – 10:00**

OTL-201 – A Clinical Study of the Investigational Haematopoietic Stem Cell Gene Therapy in MPSIIIA.

**Prof. Brian Bigger (UK)**

**10:00 – 10:30**

Combination of HSPC transplantation and cathepsin B inhibitors for treatment of Sanfilippo disease.

**Prof. Alexey Pshezhetsky (CAN)**

**10:30 – 11:00 BREAK**

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**11:00 – 11:30**

Preclinical validation of an intravenous AAV gene therapy for mucopolysaccharidosis IIIB.

**Pr. Jerome Ausseil (FR)**

**11:30 – 12:00**

JR-441 – An intravenous enzyme replacement therapy to deliver the sulfamidase enzyme to the brain to treat Sanfilippo type A.

**Dr Nicole Muschol, UKE Hamburg (DE)**

**12:00 – 12:30**

MPS IIIC Gene Replacement Therapy with scAAV9/HGSNAT Vector, JLK-247.

**Phoenix Nest (US) Speaker Jill Wood on behalf of Xin Chen**

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**12:30 LUNCH**

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#### Gene Therapy and ERT *continued*

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**14:00 – 14:30**

Investigational Therapies for Sanfilippo Syndrome and Related MPS Disorders: Denali Therapeutics' Enzyme Transport Vehicle (ETV) Platform and Clinical Updates.

**Denali Therapeutics (US)**

**14:30 – 15:00**

Long-term intracerebroventricular treatment with Tralesinidase Alfa (AX 250) for Sanfilippo type B.

**Dr Nicole Muschol, UKE Hamburg (DE)**

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**15:00 – 15:30 BREAK**

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#### Sanfilippo Symptoms and Natural History

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**15:30 – 16:00**

The role of the inflammasome and systemic inflammation in neurologic progression of Sanfilippo disease.

**Brian Bigger (UK)**

**16:00 – 16:30**

Leveraging Remote Video Capture in MPS IIIC and IIID Natural History Studies: Introducing C-RARE for Real-World Evidence to Support Clinical Trial Treatment Efficacy.

**Phoenix Nest (US) Speaker: Jill Wood**

**16:30 – 17:30**

Combining Therapeutic Approaches for Sanfilippo treatment (Participants based on attendees present)

**Scientific Round Table Discussion**

## 4TH INTERNATIONAL CONFERENCE ON SANFILIPPO SYNDROME AND RELATED DISEASES

**DAY 2 – FRIDAY, NOVEMBER 14, 2025**

### Novel Therapies and Approaches

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**08:30 – 09:00**

Harnessing the power of deep learning to repurpose drugs for the treatment of the Sanfilippo syndrome.

**Dr. Anna Fournier, Swiss Data Science Center with the Fondation Sanfilippo Suisse (CH)**

**09:00 – 09:30**

Targeting protein amyloids and brain autophagy-lysosomal pathway in Sanfilippo Syndrome and other neuronopathic MPSs.

**Alessandro Fraldi (IT)**

**09:30 – 10:00**

VRO073 – A novel molecule for the potential treatment of GM1, GM2 gangliosidosis and Gaucher Disease type 3.

**Dr Vincent Mutel, Dorphan SA (SUI)**

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**10:00 – 10:30 BREAK**

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### Screening, Biomarkers and Animal Models

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**10:30 – 11:00**

Assessment of a central nervous system demyelination in Sanfilippo disease by diffusion tensor imaging: a non-invasive method to evaluate disease progression and therapeutic response.

**Prof. Alexey Pshezhetsky (CAN)**

**11:00 – 11:30**

Biomarkers, diagnostic testing and drug development for Sanfilippo Syndrome and other MPS diseases.

**Prof. Peter Bauer, Centogene (DE)**

**11:30 – 12:00**

The effect of treatment with a heparanase-inhibiting polysaccharide on the MPS III A mouse model.

**Stéphane Sizonenko (CH)**

**12:00 – 12:45**

Family Q&A Session

Open questions and answers for families

**12:45**

Closing Remarks and Conference Wrap-up

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**13:00 LUNCH**

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