



# LET'Sanfilippo FIGHTers

*Mucopolysaccharidosis 3 Workshop*

MAY 26<sup>th</sup> - 27<sup>th</sup> 2023

## CONFERENCE PROGRAMME

📍 *Università degli studi della Campania  
"L. Vanvitelli", NAPLES (ITALY)*

Conference organized by:



**SANFILIPPO**  
Fighters

Conference venue:



Università  
degli Studi  
della Campania  
*Luigi Vanvitelli*

Registration:



# INFO AND REGISTRATION

## ABOUT THE CONFERENCE

We are delighted to announce the first Mucopolysaccharidosis 3 Italian Conference. We will welcome you in Naples from 26 to 27 May 2023 and online, around the globe. We will join forces to share knowledge, create scientific culture and advance the work to help children with Sanfilippo Syndrome. Families and caregivers, scientists and researchers, clinicians and therapists, advocates, biotechs, and supporters will be welcomed on site or online.

## JOIN ONSITE OR ONLINE

For the ultimate learning and networking experience, join us onsite in Naples. The Conference will take place at Università degli studi della Campania "L. Vanvitelli" (Via Luciano Armanni, 5 80138, Naples). If you are unable to travel, join us online.

## REGISTRATION




The Conference is free and open to everybody who cares about MPS 3. Registration is mandatory to join on site or online.

Register today at: <https://www.eubea.it/sanfilippo23/index.php> or simply scan the QR code to go directly to the registration page.

SCAN HERE →



## PROGRAMME

In the following pages, we will present the complete programme. We have reported the english and the italian talks with the icons:   We have also reported when the speaker will be remote with the icon:  ONLINE

## CONFERENCE RESOURCES

The presented abstract will be available during and after the Conference.



## CONFERENCE AIMS

The association Sanfilippo fighters was born with the purpose of helping children affected by MPSIII because we believe every child has the right to grow up and live their life to the fullest.

Since scientific research represents the only weapon with which we can fight to give Sanfilippo children a chance, the association has always put great effort in supporting scientific research, since its establishment in July 2020. In collaboration with Fondazione Telethon, Cure Sanfilippo Foundation e Sanfilippo Children's Foundation we were able to finance two research projects. The first project is focused on the pathological mechanisms involved in MPSIII, specifically on the mechanisms underlying the degeneration of the nervous tissue typical of the pathology, and the latter aid at finding an innovative treatment through the evaluation a new therapeutic approach based on the pharmacological stimulus of autophagy, the process that allows cells to dispose of waste substances with characteristically accumulate in cells in mucopolysaccharidosis.

We are also very committed to supporting families. Up to date, 35 families with affected children are part of the association. Bringing families together allow them to exchange advice, experiences and give each other support in managing their special children. We also offer them psychological support in the management of emotional stressors related not only to the management of the disease, but also to the dynamics of the relationship between parents and the siblings.

Moreover, we work very hard to raise awareness about Sanfilippo syndrome, with the hope to let as many people as possible know about this disease. Given its rarity, there are so many doctors, pediatricians, teachers and therapists who do not know it and therefore cannot recognize it in children. Understanding it is the first step in being able to deal with it.

But, probably, the most challenging work for the association is to create a network of people with different involvement in the disease. Meeting among rare families and those who closely work together with them to help their children, like clinicians and researchers, in essential to ensure the best treatments for Sanfilippo children.

For all these reasons we decided to hold this international conference to share our achievements with the scientific community and the families. We will talk about a comprehensive diagnostic related to the MPSIII disease, its physiopathological and biochemical mechanisms, the scientific projects currently in progress, the therapeutic strategies considered to date and, last but not least, we will talk about the clinical management and best care practice for Sanfilippo children.

We have a dream: finding a cure, as soon as possible! To this purpose, we are committed to work intensively and combine researchers' involvement, biotech's professionalism and families' energies.

# MUCOPOLYSACCHARIDOSIS 3 WORKSHOP

DAY 1

## OPENING

9:00 - 9:30 am	Registration
9:30 - 10:00 am	Opening and Greetings <ul style="list-style-type: none"><li>• Katia Moletta, President of Sanfilippo Fighters</li><li>• Prof. Vincenzo Nigro, University of Campania L. Vanvitelli</li><li>• Prof. Giuseppe Limongelli, University of Campania L. Vanvitelli &amp; Coordinator <i>Centro di Coordinamento Malattie Rare della Regione Campania</i></li></ul>

## TOPIC I

Pathogenesis of Sanfilippo syndrome (autophagy, GAGs and Inflammation)

### SESSION I: Autophagy-lysosomal pathways in MPS III

Chairman: Prof. Enrico Moro

10:00 – 10:05 am	Introduction to the session
10:05 – 10:25 am	<i>Drosophila melanogaster</i> models to study mitochondrial and autophagic defects in Mucopolysaccharidosis (Laura Rigon, Dipartimento di Medicina, Università degli Studi di Padova)
10:25 – 10:45 am	<i>Interplay between amyloid and autophagy in MPSIII neurodegeneration</i> (Antonio Monaco, CEINGE & University of Naples Federico II)
10:45 – 11:00 am	Coffee break




# MUCOPOLYSACCHARIDOSIS 3 WORKSHOP DAY 1

## TOPIC 1

Pathogenesis of Sanfilippo syndrome (autophagy, GAGs and Inflammation)

### SESSION II: Glycosaminoglycans and inflammation in MPS III

Chairman: Dott.ssa Laura Rigon

- 11:35 - 11:40 am** Introduction to the session
- 11:40 - 12:00 am** *Detecting the earliest proinflammatory changes and neuroinflammatory programs in Mucopolysaccharidoses: common traits may underlie shared defects between Sanfilippo and Hunter syndrome* (Enrico Moro, University of Padova)
- 12:00 - 12:20 am** *Novel dual-acting iminosugars for the treatment of Sanfilippo disease* (Luigi Pavone, University of Naples Federico II)
- 12:20 - 12:40 am** *Impairments in purine metabolic pathways in MPS III - new therapeutic targets* (K. Hemsley, Flinders University)
-  **ONLINE**
- 12:40 - 01:40 pm** **Lunch**

## TOPIC 2

Moving towards a cure for Sanfilippo Children

### SESSION III: SMALL MOLECULES THERAPIES FOR MPS III

Chairman: Dott.ssa Nicolina Cristina Sorrentino

- 01:40 - 01:45 pm** Introduction to the session
- 01:45 - 02:05 pm** *Pharmacological stimulation of brain autophagy in Sanfilippo syndrome* (Elvira De Leonibus, TIGEM/Cnr-Ibbc)
- 02:05 - 02:25 pm** *Combining high-content imaging and drug repurposing to tackle MPS type IIIA* (Diego Medina, TIGEM)
- 02:25 - 02:45 pm** *A Combined Therapy for Sanfilippo disease* (Alessandro Fraldi, CEINGE & University of Naples Federico II)
- 02:45 - 03:15 pm** **Coffee break**



# MUCOPOLYSACCHARIDOSIS 3 WORKSHOP DAY 1

## TOPIC 2

Moving towards a cure for Sanfilippo Children


### SESSION IV: Clinical Trials for Sanfilippo syndrome

Chairman: Prof. Alessandro Fraldi

- 03:15 – 03:20 pm** Introduction to the session
- 03:20 – 03:45 pm** *Overview of JCR's preclinical and clinical programs for the treatment of MPS IIIA and MPS IIIB* (Mathias Schmidt, JCR pharmaceuticals)
- 04:45 – 05:05 pm** *Development of Novel Enzyme Therapies for MPS III and other MPS disease* (Paul Harmatz, UCSF Benioff Children's Hospital)
- 05:05 – 05:25 pm**  **ONLINE** *Blood-based biomarkers for Sanfilippo syndrome* (Gal Bitan, University of California)  **ONLINE**

### SESSION I: Autophagy-lysosomal pathways in MPS III

Chairman: Prof. Enrico Moro

- 05:25 – 05:45 pm** *Sanfilippo disease type C: novel advances in pathophysiology and treatment* (Alexey Pchejetski, University of Montreal)
-  **ONLINE**

- 05:45 – 06:00 pm** **Conclusions**

# MUCOPOLYSACCHARIDOSIS 3 WORKSHOP DAY 2

## TOPIC III

Best care for children with Sanfilippo

### SESSION V: Global Clinical Management Guidelines

Chairman: Katia Moletta

<b>09:30 – 09:35 am</b>	Introduction to the session
<b>09:35 – 09:55 am</b>	<i>Sanfilippo syndrome: psychological impact on the child and the family</i> (Dr. Benedetta Greco, Ospedale Pediatrico Bambin Gesù - Roma)
<b>09:55 – 10:15 am</b>	<i>Management of patients affected by mucopolysaccharidosis</i> (Simona Fecarotta, Department of Pediatrics “Federico II”)
<b>10:15 – 10:35 am</b>	<i>Strategies to face the disease together and learn how to know it better</i> (Serena Gasperini, Fondazione IRCCS-San Gerardo dei Tintori Monza)
<b>10:35 – 10:55 am</b>	<i>The challenges of anesthetic management for MPS patients and possible solutions</i> (Simon Moser, Benioff Children’s Hospital)
<b>10:55 – 11:15 am</b>	<i>Melatonin: role in sleep physiology and therapeutic use</i> (Rodolfo Costa, University of Padova and Sara Montagnese, University of Padua & University of Surrey) <span>● ONLINE</span>
<b>11:15 – 11:45 am</b>	<b>Coffee break</b>

## TOPIC III

Best care for children with Sanfilippo

### SESSION VI: Family support

Chairman: Prof. Giuseppe Limongelli, Centro di coordinamento malattie rare Regione Campania

<b>11:45 - 11:50 am</b>	Introduction to the session
<b>11:50 – 12:10 am</b>	<i>Sibling life: Knowing and understanding who lives with a rare sibling</i> (Laura Gentile, Psicologa e psicoterapeuta, Responsabile Scientifico del Progetto Rare Sibling di OMAR and Stefania Collet, Responsabile del Progetto Rare Sibling di OMAR)

# MUCOPOLYSACCHARIDOSIS 3 WORKSHOP DAY 2

## TOPIC III

### Best care for children with Sanfilippo

- 12:10 – 12:30 am** *The assistance network for people with rare diseases and their rights (Monica Mazzucato, Centro di coordinamento malattie rare Regione Veneto)*
- 12:30 – 12:50 am** *MetabERN: the most comprehensive network of specialised centres on inherited metabolic rare diseases (Maurizio Scarpa, Centro di coordinamento regionale per le malattie rare azienda universitaria udine & MetabERN)*
- 12:50 – 01:10 pm** *MetabERN: A closer look at MetabERN's patient board community (Simon Bond, MetabERN)*
- 01:10 – 01:25 pm** **Closing remarks and final greetings**

## THE ORGANISING COMMITTEE

- Prof. Alessandro Fraldi, University of Naples Federico II & CEINGE
- Katia Moletta, Sanfilippo fighters
- Prof. Enrico Moro, University of Padova
- Dr. Angela Lombardi, Azienda Policlinico Vanvitelli and Sanfilippo Fighters
- Dr. Rosanna Aiello, Sanfilippo fighters

## THE LOCAL COMMITTEE

- Prof. Vincenzo Nigro, University of Campania L. Vanvitelli
- Prof. Giuseppe Limongelli, University of Campania L. Vanvitelli and Coordinator Centro di Coordinamento Malattie Rare della Regione Campania
- Prof.ssa Federica Papaccio, Dipartimento di Medicina, Chirurgia e Odontoiatria "Scuola Medica Salernitana", Università degli Studi di Salerno
- Prof. Michele Caraglia, University of Campania L. Vanvitelli
- Dr. Angela Lombardi, Azienda Policlinico Vanvitelli
- Dr. Anna Grimaldi, Azienda Policlinico Vanvitelli
- Dr. Carlo Caputo, University of Campania L. Vanvitelli



# MUCOPOLYSACCHARIDOSIS 3 WORKSHOP

Conference organized by:



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Fighters

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