



# NTSAD Community News

Research, Collaboration, and Community



*Supporting families  
is the center of  
everything we do...*

*October*

October 2021

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## Six Days until Imagine & Believe!

Join us as we Imagine & Believe. Be sure to **register today** for NTSAD's free, virtual Imagine & Believe event on Thursday, October 28, 7-8 PM EDT! Imagine & Believe is a celebration of the shared accomplishments of the NTSAD Community, including recent breakthroughs in research and clinical trials for individuals affected by Tay-Sachs, Canavan, GM1, and Sandhoff diseases. Families will share their perspective and hopes for the future.

We will also honor and thank Executive Director Sue Kahn for 14 years of extraordinary leadership! Join us and wish Sue farewell before she steps down next month.

**Make a Gift in Honor of Sue Kahn**

Each day leading up to the event, we are sharing on social media many of Sue's accomplishments and her lasting impact on the NTSAD community. Follow along with **#14daysofSueKahn** on **Facebook, Twitter, and LinkedIn!**

Send Sue a congratulatory message by commenting on a post or emailing a message to Sydnie Dimond at [sdimond@ntsad.org](mailto:sdimond@ntsad.org) by October 25 to have it included in a personal keepsake book especially for Sue.

*Imagine &  
Believe*

**Support Imagine & Believe**

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## Thank You Imagine & Believe Sponsors

Thank you to [Aspa Therapeutics](#), a Bridgebio Company, and all of our [Imagine & Believe sponsors](#) for their generous support and partnership. It's not too late to sponsor the event—email Susan at [skeliher@ntsad.org](mailto:skeliher@ntsad.org) for more information.

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## NTSAD Hires Next Leader, Kathleen M. Flynn

After an extensive national search, Kathy Flynn joins NTSAD as CEO following Sue Kahn's 14 years at the helm. Kathy, who starts this November, brings to our Community three decades of organizational management expertise spanning nonprofit, corporate, and academic environments. Prior to NTSAD, she served in a variety of senior leadership roles including national divisions director, executive director, community development director, and director of development at the American Liver Foundation, The Arthritis Foundation, Yale Law School, and Mercy Learning Center.



Throughout her career, Kathy has collaborated with patients, caregivers, healthcare professionals, industry members, and donors in a manner that motivated people by identifying common goals. She is a dynamic, insightful, and compassionate leader who strives to make a positive impact on people's lives by building community.

*"NTSAD plays a critical role in so many people's lives, and it is a true privilege to be the next leader of such a purposeful organization. There's never been a time of greater hope for families faced with rare diseases, and yet families need more than promise; they also need support and comfort. They need*

*an organization that they know stands up for them. That's what NTSAD is all about, and I'm thrilled to join this talented team."*

Kathy and Sue will work closely together for the next few weeks to ensure a smooth transition.

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## **Sio Gene Therapies Shares Positive Data From GM1 Clinical Trial**

Sio Gene Therapies shared positive interim safety and biomarker data from ongoing phase 1/2 clinical study of AXO-AAV-GM1 gene therapy in GM1 gangliosidosis. The FDA granted Fast Track Designation for the next steps of the investigational AXO-AAV-GM1 (AAV9-GLB1) gene therapy.

[Read the press release on positive data.](#)

[Read the press release on FDA Fast Track Designation.](#)

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## **LOTSS Think Tank Advances Therapies and Research**

Nearly 30 researchers, clinicians, industry members, and patient advocates gathered for the 4th Annual Late Onset Tay-Sachs and Sandhoff Disease (LOTSS) Think Tank, a two-day symposium designed to accelerate research toward effective treatments.

During the October event, the group discussed pre-clinical research and innovative methods, new experimental therapeutic approaches, early findings for ongoing clinical trials, the development of meaningful outcome measures, and efforts to establish newborn screening for GM2 as well as GM1 and Canavan disease.

Keynote speaker Jeremy Schmahmann, MD, Massachusetts General Hospital and Harvard Medical School, shared a new perspective on the brain's cerebellar function known for coordinating movement, but it may also regulate cognition and emotional function. Throughout his speech, he emphasized the importance of listening to patients and having them be at the very center of the therapeutic process.

The Think Tank was sparked by Alexis Buryk and was inspired by her twin daughters, who are living with LOTSS. Together the family has raised more than \$700,000 for LOTSS research via the [Katie & Allie Buryk Fund](#), and they are sponsors of The Think Tank. NTSAD and National Human Genome Research Institute (NHGRI) help organize the event and steward its ongoing priorities. Special thanks to Cynthia Tiff MD, PhD (NHGRI) and Steve Walkley DVM, PhD (Albert Einstein College of Medicine) for organizing and co-chairing the meeting.



## NTSAD Awards Grant to Sustain Tay-Sachs Sheep



NTSAD has awarded a grant to Heather Gray-Edwards, PhD at the University of Massachusetts Medical School (UMMS) to sustain the flock of Tay-Sachs sheep for the next year for researchers as this sheep model is considered to be the most similar to the Late Onset form of Tay-Sachs in humans.

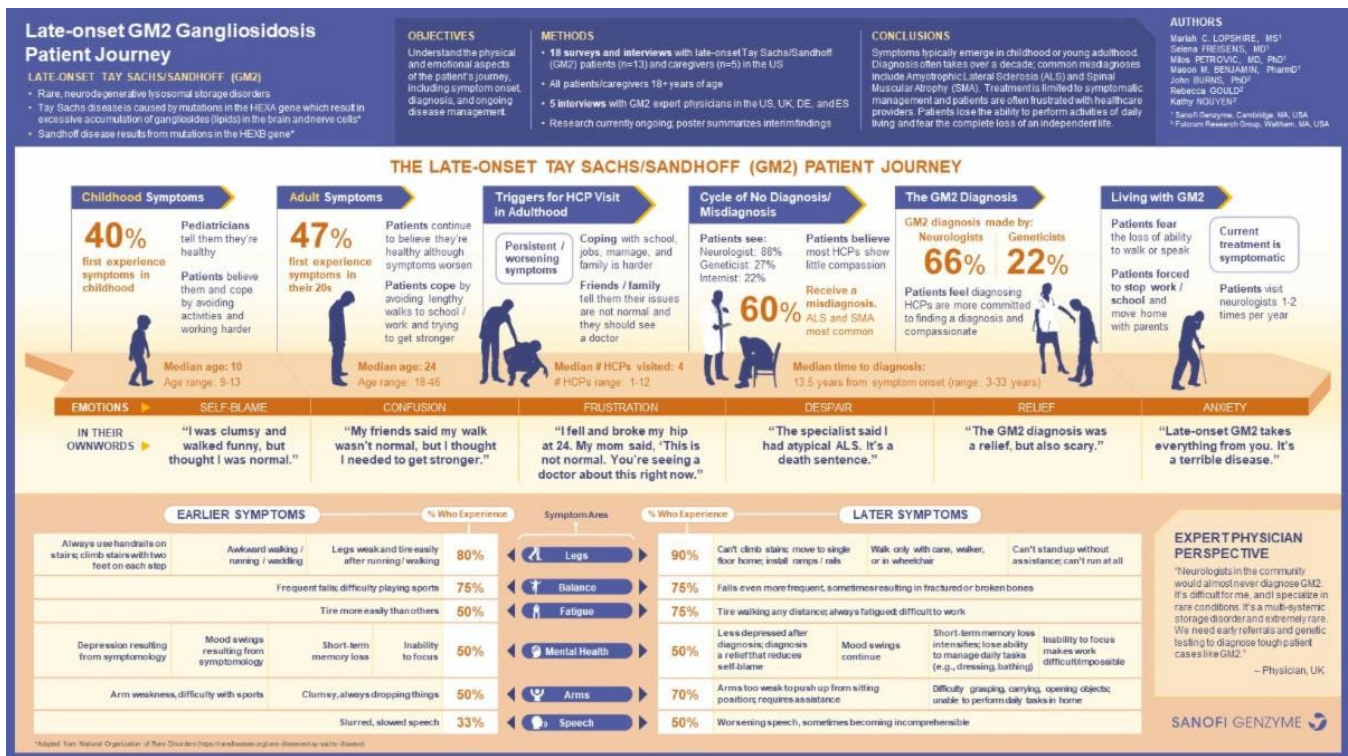
Animal models that mimic human diseases are essential at all stages of research but are often difficult and expensive to create. Large animals, such as the Tay-Sachs Disease sheep, are particularly valuable for preclinical work that ensures the safety of a treatment or a drug. Therefore, maintaining the TSD sheep flock is important to enable future research towards developing a cure for Tay-Sachs and Sandhoff diseases.

During the last 12 years, NTSAD has invested more than \$400,000 toward research involving the TSD sheep model, including maintenance when the flock was first identified at the Horaks' farm in Texas, and then when they were housed at Auburn University. Previously, NTSAD made a grant to Doug Martin PhD and Auburn University to prove safety and efficacy in the Tay-Sachs sheep model. Later, after Dr. Gray-Edwards and the sheep relocated to UMass Medical School, NTSAD awarded another grant to determine the efficacy of adeno associated viral (AAV) gene therapy after cerebrospinal fluid (CSF) delivery in the TSD sheep.

Thank you [Katie & Allie Buryk Research Fund](#), [the Heringer Fund](#), the LOTS Research & Educational Foundation, and [Vera's Fund](#) for support to sustain the flock of sheep.

## Late Onset Community's Journey to Diagnosis

At [NORD's](#) recent Breakthrough Summit, Sanofi Genzyme presented the company's poster illustrating the often-arduous diagnostic journey of patients, caregivers, and families affected by Late-Onset GM2 Gangliosidosis. As the Summit is the largest gathering of key stakeholders in the rare disease community, the information heightens awareness of the heterogeneity of the disease and will help accelerate future diagnosis.



## Day of Hope 2021

Through the extraordinary efforts of NTSAD's community members, their friends, and families, the 2021 Day of Hope raised more than \$50,000 for research thus far.

Thank you to the more than 20 families who participated by creating t-shirt campaigns, sharing NTSAD's Rare Aware infographics and social media posts, encouraging donations from friends and families, and most of all for hosting personal events in their communities.

We also thank all industry partners who demonstrated they #Care4Rare on social media.



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## The Rudness Family Raises Rare Awareness

Thank you to Kim and Adam Rudness for spreading awareness of Tay-Sachs disease by sharing their son Greyson's story. In the interview with their local news station in Michigan, they highlight the importance of carrier screening for rare diseases. Kim and Adam hope to inspire other couples in their community and beyond to get tested before starting a family.

**[Watch it now!](#)**



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**NTSAD leads the worldwide fight to treat and cure Tay-Sachs, Canavan, GM1, and Sandhoff diseases by driving research, forging collaboration, and fostering community. Supporting families is the center of everything we do.**

**Donate**

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### STAFF

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