



# NTSAD Community News

Research, Collaboration, and Community



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

February 2021

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## Sio Gene Therapies Doses First GM2 Patient

**Sio Gene Therapies** dosed the first patient in the Phase 1/2 trial for the company's investigational gene therapy, AXO-AAV-GM2, for the treatment of patients with GM2 gangliosidosis.

[Read the Press Release.](#)

## Lysogene Prepares Start of GM1 Clinical Trial

**Lysogene** receives permission from the FDA to start the clinical trial of gene therapy in the U.S. with LYS-GM101 in the treatment of GM1 gangliosidosis. Trial enrollment expected to begin in first six months of 2021.

Lysogene previously received approval for LYS-GM101 in the U.K.

[Read the Press Release.](#)

## Sheryl Sandberg Talks Resiliency with NTSAD Community

**Sheryl Sandberg, Facebook COO and co-author of *Option B: Facing Adversity, Building Resilience, and Finding Joy*, will discuss the challenges and emotions experienced when facing life-altering, unexpected situations on February 22 at 7 p.m. EST.** As part of NTSAD's Choosing Resiliency in the Face of Adversity Family Connection Webinar Series, Sheryl will be joined by Blyth Taylor Lord, founder of Courageous Parents Network and NTSAD Board member and moderator, Becky Benson, NTSAD's Family Services and Conference Coordinator. The event is free to attend, but registration is required.

[Register Now for the Webinar.](#)

## Attend and Sponsor NTSAD's Annual Family Conference

**Join us on April 22-25, 2021 for the 43rd Annual Family Conference and the second, virtual conference.** Whether virtual or in person, NTSAD provides families with a safe space to support and learn from one another, to gain knowledge directly from researchers and industry working on potential therapies, and to contribute and share their experiences as our community navigates the launch of several clinical trials.

In 2020, the virtual conference was our largest ever with more than 400 registrants, including nearly 150 families participating from across the U.S., Canada, and ten other countries.

During the conference families receive incredible support from one another and a sense of belonging to a community. The conference is very powerful experience. Many families participate every year to connect with friends, who have become like family, and to support and mentor new families. One mother, after attending her first conference shared,

***“You don’t know true grace until you meet a mother holding her dying child in her arms reaching out to you, a total stranger, with love and compassion. I will never be the same.”***

The NTSAD Annual Family Conference is made possible by the generosity of industry partners, foundations, families and individuals. Sponsorship goes toward family programming and support as well as sustaining NTSAD’s family services. NTSAD incurs expenses for event hosting and production, software, speaker fees, and supplies, including a conference gift box with activities for NTSAD families and individuals living in the U.S.

**View sponsorship levels.**

**Read more about this year’s conference.**

**The conference is free to attend, and registration for industry members, researchers, clinicians begins in early March.**

To sponsor the conference email NTSAD’s Director of Development and Communications Susan Keliher at [skeliher@ntsad.org](mailto:skeliher@ntsad.org).



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## Ways to Show You Care for Rare



Rare Disease Day is February 28, and there are numerous activities, events, and advocacy opportunities to help shine a light on rare disease patients and their families as well as researchers and clinicians--all working together in leading the fight for effective treatments.

Check out **the National Organization for Rare Disorders (NORD) website** to read rare patient stories, sign up for virtual events, and find resources to help you or your company [#showyourstripes](#) on social media!

Connect with the Rare Community through the official **Rare Disease Day website**, and share your rare stories and photos. Watch the official **Rare Disease Day 2021 video** and meet six heroes from six continents all living with a rare disease.

Watch B Brave Foundation's **Project Rare Docuseries**, which offers six episodes highlighting families, caregivers, and healthcare professionals navigating the complexities of grief, clinical trials, and caring for loved ones with rare, incurable diseases during the COVID-19 crisis.

Attend National Institute of Health's **Rare Disease Day Conference** on March 1 from 5:30 p.m. EST. This year's event will feature interactive panel discussions, rare stories through TED-style talks, and more. The conference is free and open to the public.

Wear that you care by purchasing an NTSAD Rare Bear T-shirt. There is version for everyone in the NTSAD Community--Rare Bear, Mama Bear, Papa Bear, Grand Bear, Bro, Sis, and Rare Support Bear. The t-shirts come in coordinating colors and various sizes for your Rare Bear family or Rare colleagues. **Order your t-shirt today** and support NTSAD at the same time. A portion of proceeds go toward NTSAD.

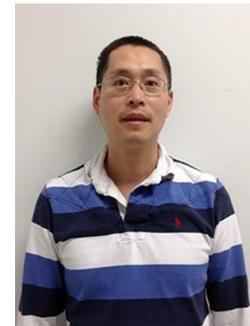


Engage and share NTSAD's own Rare Disease Day Social Media posts on highlighting NTSAD's Rare Families!

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## NTSAD-funded Researcher Identifies Biomarker to Assess GM1 Therapies

In 2018, NTSAD funded a research investigation of biomarker H3N2b led by Xuntian Jiang, PhD, Washington University School of Medicine. The project has been completed and results have shown that H3N2b is a good biomarker for treatment in GM1 patients undergoing gene therapy.



A major challenge for developing treatments for GM1 is the difficulty in the evaluation of efficacy, particularly due to limited patient numbers and heterogeneity in age, severity of symptoms, and stage of disease progression. Biomarkers that reflect disease status could provide a valuable tool for assessing the effect of treatment and reasonably predict its clinical benefit.

Research has identified a natural component, an oligosaccharide (or a carbohydrate whose molecule is composed of a relatively small number of monosaccharides), in the biomarker H3N2b that is significantly elevated in the urine, cerebrospinal fluid (CSF), and plasma from GM1 patients as well as in brains of animals, specifically from the GM1 cat model.

The research found the H3N2b in GM1 cat brains was inversely correlated with  $\beta$ -galactosidase enzyme activity, an indicator of GM1, which is reduced following gene therapy. **Thus, the results find that H3N2b is a good biomarker for the assessment of gene therapy treatment efficacy in GM1 patients.**

Based on preliminary results of the project, **an NIH U01 grant application was filed and funding was approved** to complete the method validation of the H3N2b in plasma and CSF and also use the validated method to assess AAV gene therapy treatment efficacy in the clinical trial over five years. It may prove to be a valuable biomarker for other GM1 therapies, as well.

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## NTSAD to Hire Research Director

NTSAD is seeking to hire the organization's first Research Director to engage key stakeholders regarding the discovery and development of treatments, diagnostics, and technologies for Tay-Sachs, Canavan, GM1 gangliosidosis, and Sandhoff diseases. Consider joining NTSAD -- leading the fight since 1957!

**Learn More and Apply Here.**

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## **Sena-Esteves Shares the Journey of Gene Therapy for Tay-Sachs**

At Quinnipiac University's Annual Rare Disease Day Symposium, Dr. Miguel Sena-Esteves, associate professor of neurology at the University of Massachusetts Medical School, will share his expertise and experience in the development of AAV gene therapy for Tay-Sachs Disease. His address, From Bench-to-Bedside through the Valley of Desperation, will take place on February 27 at 10 a.m. EST. In addition to conducting the first in-human clinical trial of AAV gene therapy for Tay-Sachs disease, Dr. Sena-Esteves is leading the pre-clinical development efforts for a GM1-gangliosidosis AAV gene therapy in collaboration with Lysogene.



**Learn More about the Symposium.**

**Register for Dr. Sena-Esteves Session**

**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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